THE 10th EUROPEAN CONFERENCE ON RARE DISEASES & ORPHAN PRODUCTS

ecrd

ONLINE on 14-15 May 2020

THE JOURNEY OF LIVING WITH a RARE DISEASE in 2030

ONLINE PROGRAMME

Organised by

EURORDIS RARE DISEASES EUROPE

Co-organised by

orphanet Inserm

With the support of

AFMTÉLÉTHON INNOVER POUR GUÉRIR

Co-funded by the Health Programme of the European Union

#ECRD2020

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All times listed are Central European Summer Time (CEST)

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**

Prof. Milan Macek
Professor of Medical and Molecular Genetics, Motol University Hospital and Charles University Prague, Czech Republic

Maria Montefusco
President,
Rare Diseases Sweden

Violeta Stoyanova-Beninska
Chair, Committee of Orphan Medical Products, European Medicines Agency

Dear Participants,

We are delighted to welcome you on behalf of conference organisers EURORDIS-Rare Diseases Europe, co-organisers Orphanet, Rare2030 project partners, and all other conference partners to the 10th European Conference on Rare Diseases & Orphan Products 2020. Globally recognised as the largest, patient-led rare disease event, this is the first time the conference is **being held exclusively online!**

In these unprecedented times, we need to **come together to shape a better future for people living with a rare disease**. Rare diseases should not be forgotten in the current pandemic, since they also represent a vulnerable group of citizens. The COVID-19 pandemic has exposed the cracks in healthcare systems worldwide and has demonstrated that a single country cannot manage such complex issues alone. The pandemic has further substantiated the importance of the EU wide cross-border collaboration on healthcare. This is the time to deepen our commitment, strengthen our ties and to learn from each other in the face of adversity.

As the EU shapes its future policy and spending frameworks for the coming decade, ECRD 2020 serves as an opportunity to already **co-design policy options today** that can lead to better conditions for people living with rare diseases for the years ahead. This is why we felt it vital to maintain the event online and **do our best, together with you, to turn this crisis into an opportunity to drive change**.

We are therefore delighted that you are joining ECRD 2020 online to take part in discussions on how to build the future ecosystem of policies and services needed to drive this change. **We very much value your contribution, your expertise and your opinions. You have a unique opportunity to "Be the future you want"!**

The health of 30 million people living with a rare disease in Europe should not be left to chance. The ECRD 2020 theme "The journey of living with a rare disease in 2030" recognises that the next decade holds great potential for improvement and that while we cannot predict the future, we all have a key role in preparing for it. Now isn't the time to sit on the side-lines. Get involved and help ensure that people living with a rare disease have the right to reach their highest potential of well-being.

We hope you enjoy ECRD 2020 as **an unrivalled opportunity to network** and exchange invaluable knowledge with **all stakeholders in the rare disease community** from over 40 countries around the world - patient representatives, policy makers, researchers, clinicians, industry representatives, payers and regulators.

We invite you to take a glance at the full programme, view the high quality posters on display, take a virtual walk around the exhibit hall and compete with fellow participants to win a prize on our interactivity leader board!

We sincerely hope you enjoy ECRD 2020 online,

ECRD 2020 Programme Committee Co-Chairs

PROGRAMME AT A GLANCE

All times listed are Central European Summer Time (CEST)

Thursday 14 May 2020

10:30 - 11:30	Virtual Platform Navigation & Online Networking					
11:30 - 13:00	Opening & Plenary Session					
13:00 - 14:00	Lunch & Browsing Posters, Exhibit Hall and Networking					
13:00 - 14:00	COVID-19: a time for exchange					
14:00-14:45	Plenary Session: Rare2030 Foresight scenarios					
Themes	1/ The future of diagnosis: new hopes, promises and challenges	2/ Our values, our rights, our future: shifting paradigms towards inclusion	3/ Share, Care, Cure: Transforming care for rare diseases by 2030	4/ When therapies meet the needs: enabling a patient-centric approach to therapeutic development	5/ Achieving the triple As by 2030: Accessible, Available and Affordable Treatments for people living with a rare disease	6/ The digital health revolution: hype vs. reality
14:45-16:15	Session 0101	Session 0201	Session 0301	Session 0401	Session 0501	Session 0601
16:15 - 16:45	Comfort Break & Browsing Posters, Exhibit Hall and Networking					
16:45-18:15	Session 0102	Session 0202	Session 0302	Session 0402	Session 0502	Session 0602
18:15 - 19:30	Global Commission to End the Diagnostic Odyssey for Children with a Rare Disease: Progress Update					
18.30 - 19.30	Meet the Speakers, Speed Networking, Browsing Posters & Exhibit Hall					

Friday 15 May 2020

10:00-11:00	Plenary Session & Poster Winners Presentations					
11:00-11:30	Comfort Break & Browsing Posters, Exhibit Hall and Networking					
Themes	1/ The future of diagnosis: new hopes, promises and challenges	2/ Our values, our rights, our future: shifting paradigms towards inclusion	3/ Share, Care, Cure: Transforming care for rare diseases by 2030	4/ When therapies meet the needs: enabling a patient-centric approach to therapeutic development	5/ Achieving the triple As by 2030: Accessible, Available and Affordable Treatments for people living with a rare disease	6/ The digital health revolution: hype vs. reality
11:30 - 13:00	Session 0103	Session 0203	Session 0303	Session 0403	Session 0503	Session 0603
13:00 - 14:00	Lunch Break & Browsing Posters, Exhibit Hall and Networking					
13:00 - 14:00	COVID-19: a time for exchange					
14:00-15:30	Session 0104	Session 0204	Session 0304	Session 0404	Session 0504	Session 0604
15:30 - 16:00	Comfort Break & Browsing Posters, Exhibit Hall and Networking					
16:00 - 18:00	Closing Plenary Session					
18:00 - 19:00	Meet the Speakers, Speed Networking, Browsing Posters & Exhibit Hall					

COMMITTEES

PROGRAMME **COMMITTEE**

Diego Ardigo
Chiesi Farmaceutici

Dimitrios Athanasiou
World Duchenne Organisation

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Ana Rath
Orphanet

Andrea Ricci
ISINNOVA

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CEO, Genetic Alliance UK

Birute Tumiene
Institute of Biomedical Sciences, Vilnius University

COMMITTEES

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EURORDIS

Ines Hernando
EURORDIS

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EURORDIS

Denis Costello
CML Advocates Network

Maria Cavaller
EURORDIS

Simone Boselli
EURORDIS

Gulcin Gumus
EURORDIS

OUTREACH **COMMITTEE**

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Finnish Network for Rare Diseases

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Genetic Alliance UK

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Rare Diseases Italy (UNIAMO)

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Rare Diseases Denmark (Sjaeldne Diagnoser)

Ingeborg Vea
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Association for Children Rare Diseases

Mirjam Mann
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Miroslaw Zielinski
ORPHAN

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Martina Michalova
Rare diseases Czech Republic

Baiba Ziemele
Latvian Alliance for Rare Diseases
VSOP

Helga Gruden
EINSTÖK BÖRN

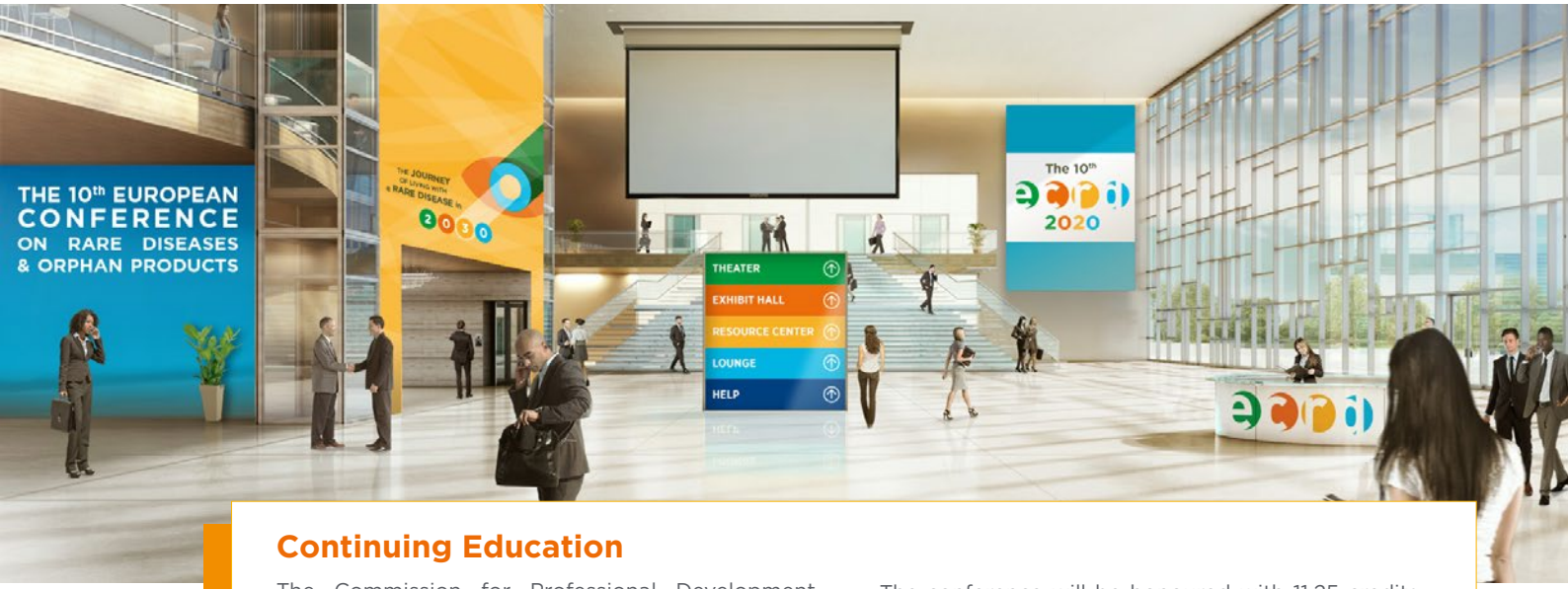
Cor Oosterwijk
VSOP, Netherlands

A VIRTUAL CONFERENCE

EURORDIS has selected Intrado as the online, virtual conference platform through which the ECRD 2020 will take place. Intrado has successfully executed over 40,000 virtual events and webcasts annually in over 157 countries, helping organisations achieve success by streamlining the way they communicate through virtual events.

FEATURES OF THE VIRTUAL CONFERENCE

- Delegates can view in real time and **switch between parallel sessions** and **access recordings of all sessions** for up to one year after the conference within the platform.
- The online platform is being built to guarantee **online networking opportunities** with speakers and fellow conference delegates.
- **Posters and a virtual exhibit** hall are integrated.
- Compete with fellow participants to **win a prize** on our interactivity leader board, making connecting from your home or office more fun!



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 11.25 credits for pharmaceutical medicine.

SwAPP
Swiss Association of Pharmaceutical Professionals

VIRTUAL TOUR OF THE PLATFORM

Join us for a pre-conference Facebook live virtual tour of the online platform on 13 May from 14.30 – 15.30 CEST!

https://www.facebook.com/events/677443599706976/

www.rare-diseases.eu/virtualtour

The online platform for the conference will be live from 11.30am CEST on Wednesday, 13 May to give you a chance to familiarise yourself with it, to ensure you get the best experience out of ECRD 2020 sessions and networking!

INTERPRETATION AND SUBTITLES

Interpretation of the following sessions will be available via the Interprefy app from English into French & German:

Day 1: 14 May

Opening Plenary session: 11.30 – 13.00 CEST

Rare2030 Plenary session: 14.00 – 14.45 CEST

Day 2: 15 May

Closing Plenary session: 16.00 – 18.00 CEST

Connection instructions

An automated subtitling/captioning function will be available within the platform for all sessions

IN PARTNERSHIP WITH

With co-funding from

AFMTÉLÉTHON
INNOVER POUR GUÉRIR

Co-funded by
the Health Programme
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ÅGRENSKA

DIA

efpia
European Federation of Pharmaceutical Industries and Associations

EUROPEAN JOINT PROGRAMME
RARE DISEASES

ESHG
EUROPEAN SOCIETY OF HUMAN GENETICS

EUCOPE
European Confederation of
Pharmaceutical Entrepreneurs AISBL

EuropaBio®
The European Association for Bioindustries

hope
European Hospital and
Healthcare Federation

MedTech Europe
from diagnosis to cure

NGO COMMITTEE FOR
RARE DISEASES

Rare2030
Foresight in Rare Disease Policy

RARE
DISEASES
INTERNATIONAL

SÄLLSYNTA
DIAGNOSER
RARE DISEASES SWEDEN

ASSOCIATE PARTNERS

European Reference Networks

European Reference Network | Bone Disorders (ERN BOND)

Erasmus MC Universitair Medisch Centrum Rotterdam

European Reference Network for rare or low prevalence complex diseases
Network Endocrine Conditions (Endo-ERN)

Epi CARE

ERKNet The European Rare Kidney Disease Reference Network

European Reference Network for rare or low prevalence complex diseases
Network Inherited and Congenital Anomalies (ERNICA)

European Reference Network | Adult solid Cancers

European Reference Network | Hematological Diseases (ERN EuroBloodNet)

European Reference Network | eUROGEN *Urogenital Diseases*

European Reference Network ERN-EYE

European Reference Network | Genetic Tumour Risk Syndromes (ERN GENTURIS)

European Reference Network for rare or low prevalence complex diseases
Network Heart Diseases (ERN GUARD-HEART)

ITHACA European Reference Network

European Reference Networks ERN-LUNG RARE RESPIRATORY DISEASES

European Reference Network | MetabERN European Reference Network for Hereditary Metabolic Disorders

European Reference Network for rare or low prevalence complex diseases
Network Hepatological Diseases (ERN RARE-LIVER)

European Reference Network | Connective Tissue and Musculoskeletal Diseases (ERN ReCONNET)

European Reference Network for rare or low prevalence complex diseases
Network Neurological Diseases (ERN-RND)

European Reference Networks | SKIN

Transplantchild

European Reference Network for rare or low prevalence complex diseases
Network Transplantation in Children (ERN TRANSPLANT-CHILD)

European Reference Network for rare or low prevalence complex diseases
Network Vascular Diseases (VASCERN)

ASSOCIATE PARTNERS

ASSOCIATE PARTNERS

NETWORKING EVENTS

#ECRD2020

The online conference platform has been designed to guarantee a truly immersive and virtual experience for all participants. You will be able to network with exhibitors directly on the virtual booths and with fellow delegates using the intra-environment emails or instant chats along with open discussions in the lounge area with a special COVID-19 group chat included.

Free time to visit the exhibition and posters has been integrated into the programme along with the following facilitated networking opportunities with speakers and fellow conference delegates:

DAY 1: 14 MAY

13.00 – 14.00 CEST

COVID-19 – a time for exchange:

Results of the EURORDIS COVID-19 survey to be presented followed by an open exchange in smaller groups about how the pandemic is impacting your **healthcare and treatment**. Go to the group discussions via the 'Sessions' section of the platform to join this group.

18.30 – 19.30 CEST

Meet the Speakers:

Take this opportunity to ask additional questions to a selected number of speakers from Day 1 in 6 parallel discussion rooms split according to the six conference themes. Go to the group discussions via the 'Sessions' section of the platform to join these rooms.

18.15 – 19.30 CEST

Speed networking:

Be matched at random with fellow delegates keen to re-create the corridor conversations that take place by chance at conferences. A series of small breakout groups will be created during this session to be sure you meet as many people as possible. Get your aperitif ready! Go to the group discussions via the 'Sessions' section of the platform to join these rooms.

DAY 2: 15 MAY

13.00 – 14.00 CEST

COVID-19 – a time for exchange:

Results of the EURORDIS COVID-19 survey to be presented followed by an open exchange in smaller groups about how the pandemic is impacting **employment and access to social services**. Go to the group discussions via the 'Sessions' section of the platform to join this group.

18.00 – 19.00 CEST

Meet the Speakers:

Take this opportunity to ask additional questions to a selected number of speakers from Day 2 in 6 parallel discussion rooms split according to the six conference themes. Go to the group discussions via the 'Sessions' section of the platform to join these rooms.

18.00 – 19.00 CEST

Speed networking:

What better way to end the conference than taking part in a networking roulette where you will be matched at random with fellow delegates keen to re-create the corridor conversations that take place by chance at conferences. A series of small breakout groups will be created during this session to be sure you meet as many people as possible. Get your aperitif ready! Go to the group discussions via the 'Sessions' section of the platform to join these rooms.

OPENING AND PLENARY SESSION AGENDA

Thursday, 14 May 2020 - 11:30 - 13:00

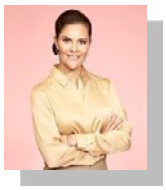
The Opening and Plenary Sessions will be simultaneously interpreted from English into 2 languages:

French
German

MODERATOR

Lise Murphy, Patient Advocate, Marfanföreningen (Swedish Marfan Association)

GREETINGS



HRH Crown Princess Victoria of Sweden

Photo: Anna-Lena Ahlström, The Royal Court of Sweden

WELCOME ADDRESS & OPENING REMARKS

Terkel Andersen, President, EURORDIS-Rare Diseases Europe

Ana Rath, Director, Orphanet

Maria Montefusco, President, Rare Diseases Sweden

KEYNOTE ADDRESSES

Lena Hallengren, Swedish Minister of Health & Social Affairs, Sweden

Stella Kyriakides, European Commissioner for Health and Food Safety

Helena Dalli, European Commissioner for Equality, Malta

Irene Norstedt, Director in DG Research & Innovation of the European Commission (Statement by Mariya Gabriel, Commissioner for Innovation, Research, Culture, Education & Youth, Bulgaria)

INSPIRATIONAL SPEAKER

'Love, Support, Encouragement and Demands – Being a Policy Maker with a Rare Disease'

David Lega, Member of the European Parliament, Sweden

PLENARY SESSION

Thursday, 14 May 2020 - 14:00 - 14:45

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

French
German

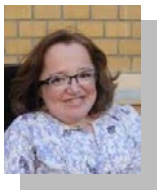
RARE 2030 FORESIGHT SCENARIOS

Rare 2030 Overview

Prof. Milan Macek, Motol University Hospital and Charles University, Prague

Video: Rare 2030 "What If" Scenarios

Personal Perspective



Rebecca Skarberg, Osteogenesis Imperfecta Federation Europe (OIFE), Norway

Audience Voting

PLENARY SESSION

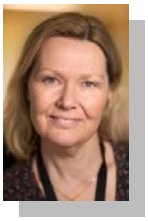
Friday, 15 May 2020 - 10:00 - 11:00

MODERATOR

Lise Murphy, Patient Advocate, Marfanföreningen (Swedish Marfan Association)

KEYNOTE ADDRESSES

'Discoveries for the Benefit of Man: Lessons from the Past and Hope for the Future'



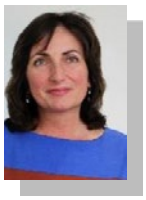
Professor Anna Wedell, Member and Former Chair of the Nobel Committee for Physiology or Medicine, Sweden

'Life languages and red flags in the red sand'

Dr Gareth Baynam, Clinical Geneticist, Genetic Services of Western Australia

POSTER WINNERS

Introduced by:



Dr. Violeta Stoyanova-Beninska, Chair, Committee for Orphan Medicinal Products, EMA

1st Place: P 163 - A collaborative and patient-centric effort to find the first effective treatment for alkaptonuria. (**Nick Sireau**)

2nd Place: P 267 - 'Patient Journeys': Personal experiences shaping clinical priorities (**Olivia Spivack**)

3rd Place: P 268 - Quality of life (QoL) for people with rare diseases: Recruitment challenges and consequences in a study pilot-testing the UK-PSC-QoL, a provisional QoL tool for people with primary sclerosing cholangitis (PSC) in the UK (**Elena Marcus**)

THEME 1

THE FUTURE OF DIAGNOSIS: NEW HOPES, PROMISES AND CHALLENGES

THEME LEADERS:

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

Virginie Bros-Facer, Scientific Director, EURORDIS

THEME SUPPORT :

Gulcin Gumus, Research and Policy Project Manager, EURORDIS

THEME DESCRIPTION:

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

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

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

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

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

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

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

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

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

Speakers:

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

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

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

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

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

Newborn Screening: Now and in the Future

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

Chair: Jayne Spink, CEO, Genetic Alliance UK

Speakers:

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

Nick Meade, Director of Policy, Genetic Alliance UK

Sara Hunt, Chief Executive Officer, Alex TLC

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

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

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

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

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

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

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

Speakers:

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

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

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

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

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

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

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

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

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

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

This session will explore what happens after the completion of genetic analysis from the perspective of patients and health professionals. The session aims to explore what is important to patients regarding their potential diagnosis (or lack thereof) and future care pathways; to discuss innovative ways of working with health professionals and patients to develop interventions which support these care pathways; and to appreciate the developing role of networks in the delivery of new approaches to aspects of care for rare diseases

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

Speakers:

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

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

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

Dr. Vera Frankova, Univerzita Karlova

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

THEME 2

OUR VALUES, OUR RIGHTS, OUR FUTURE: SHIFTING PARADIGMS TOWARDS INCLUSION

THEME LEADERS:

Maria Montefusco, President, Rare Diseases Sweden, Sweden

Valentina Bottarelli, Public Affairs Director and Head of European and International Affairs, EURORDIS

THEME SUPPORT :

Clara Hervas, Public Affairs Manager, European and International Affairs, EURORDIS

THEME DESCRIPTION:

Evidence demonstrates that people living with a rare disease and their families continue to face serious every day and social inclusion challenges. Rare diseases ensue in a high level of psychological, social and economic vulnerability and are detrimental to people's active participation in society.

This interlinkage between rarity, vulnerability, inequalities and social exclusion means that there is space for the integration of rare disease strategies into the broader human rights' agenda, and the health and development efforts at global level, with clear opportunities to contribute to the United Nations Agenda 2030: the Sustainable Development Goals (SDGs). This goal-based framework was agreed in 2015 by all UN Member States with the clear principle to "leave no one behind". In this sense, the SDGs are interdependent, universal goals that aim to address global challenges (such as poverty, health and climate).

The synergies between the SDGs and rare diseases have been acknowledged in different events, reports and texts of the UN, and there is momentum for international commitments for the benefit of persons living with a rare disease. In particular, the rare disease community has voiced the need to be included in efforts to achieve Universal Health Coverage (UHC) (as part of SDG 3 which focuses on health), as well as in efforts to ensure non-discrimination on the basis of health or disability status (as part of SDG 10 on reducing inequalities, or SDG 8 on decent work for example).

This theme will look at these ongoing advances in international advocacy and discuss the significance they have for different stakeholders of the rare disease community. A key goal of the theme will be to discuss how the rare disease community can translate those global commitments and aspirations into concrete regulatory practices and policies in the national context, which will have a real impact on the daily lives of those affected. Ongoing trends like reduced funding for human rights, an increasingly challenging political and social space, a debilitation of the values of solidarity and equity, and a shrinking space for civil society will be part of the debate, but the overall objective of the theme will be to identify the best practices and potential new practices that prove the value of investing in human rights and inclusion.

PRE-RECORDED SESSION: (Available on demand)

Getting our rights 'right': An international framework for rare diseases

Why should European citizens be concerned by the decisions being taken at the United Nations? Why should international collaboration continue to be fostered in the field of rare diseases? What sort of traction and impact can international frameworks have at the national level? To explore these questions, this session will first frame rare diseases as a policy priority, showcasing the EU approach and its framework established for addressing this issue. The session will then move on to frame rare diseases within the global human rights and Sustainable Development Goals (SDGs) agendas and present the advances made so far in international advocacy in engaging the United Nations bodies and agencies (UN General Assembly, Office of the High Commissioner for Human Rights, Human Rights Council…). Finally, the session will explore how to continue the progress towards a global t agenda that addresses the needs of persons living with a rare disease and encourages enforcement nationally, through tools such as UN resolutions.

Chair: Yann Le Cam, Chief Executive Officer, EURORDIS-Rare Diseases Europe

Speakers:

Anders Olauson, Chair, NGO Committee for Rare Diseases

Dr. Durhane Wong-Rieger, Canadian Organization for Rare Disorders

Raquel Peck, Senior Advisor and Former CEO of World Hepatitis Alliance, Switzerland

Todd Howland, Chief of the Development and Economic and Social Issues Branch, UN OHCHR

SESSION 0201: Thursday 14 May 2020, 14:45 – 16:15

UHC: from political commitment to reality for all

What is the value of Universal Health Coverage? What will the impact be in Europe: in terms of population, of services provided and of percentage of coverage? How will the EU implement the international commitments on UHC? Do national strategies on rare diseases successfully ensure the rights to health of persons living with a rare disease? How are patient's rights in cross-border care implemented across Europe and do they actually ensure access to health when this is not provided at country level? This session will look at these sorts of questions during a panel discussion on our current system and whether it may be outdated and in need of a paradigm shift. Possible trends like increased social investment, early intervention and prevention and promotion and the consequences of this on the balance between primary and secondary care will be part of the discussion.

Chair: Dr. Nata Menabde, Executive Director, WHO Office at the UN

Speakers:

Dr Suvanand Sahu, Deputy Executive Director, Stop TB Partnership Secretariat

Matt B. Johnson, Rare Diseases International Healthcare Advisor, WHO Collaborative Global Network for Rare Diseases, Germany

Martin Seychell, Deputy Director-General for Health and Food Safety, DG SANTE, European Commission, Belgium

Vytenis Andriukaitis, WHO Special Envoy for the European Region, Lithuania

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SESSION 0202: Thursday 14 May 2020, 16:45 – 18:15

Holistic Care for People Living with Rare Diseases: The Future is Now

"The specific nature of rare diseases also calls for a holistic, comprehensive and multi-disciplinary response, deeply grounded by essence in a human rights vision" (Dainius Puras, UN Special Rapporteur on the right of everyone to the enjoyment of the highest attainable standard of health). This session looks at what holistic care means in practice for persons living with a rare disease from a human rights framework and a societal point of view, rather than solely in terms of provision. It will particularly focus on the opportunities that holistic care offers, but will also look at the risks or hurdles that may be encountered to fully implement this. The session could also be an opportunity to look at how rights are interpreted in different scenarios, and therefore enshrined into and enforced (or not) within different models of care, which may depend on the direction that society as a whole chooses to take.

Chair: Robert Hejdenberg, CEO, Agrenska, Sweden

Speakers:

Ann Nordgren MD, PhD, professor in Clinical genetics at the Karolinska Institute and senior consultant at the Karolinska University hospital

Rebecca Tvedt Skarberg, Advisor, Osteogenesis Imperfecta Federation Europe (OIFE), Norway

Dr. Encarna Guillén, Head of Unit of Medical Genetics, Paediatric ward, Hospital Clínico Universitario Virgen de la Arrixaca, Spain

Dr. Cecilia Gunnarsson, Division of Clinical Genetics, Department of Clinical and Experimental Medicine, University Hospital, Linköping University, Linköping, Sweden

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SESSION 0203: Friday 15th May 2020, 11:30 – 13:00

Rare and Equal: Ensuring Non-Discrimination on the Basis of Health and Disability

People living with a rare disease often face discrimination and stigma based on their health status or chronic condition, experiencing unequal treatment in a number of areas ranging from access to education, employment, leisure and other essential support services. This is aggravated when the illness is invisible or changing. This session will present the major figures on this issue as well as personal stories and will look at the ways of enforcing these persons' rights (CRPD articles 5, EU Charter of Fundamental Rights,

European Pillar of Social Rights), particularly by ensuring appropriate regulatory processes and making use of existing tools.

Chair: Maria Montefusco, President, Rare Diseases Sweden

Speakers:

David Lega, Member of the European Parliament (MEP), Sweden

Jana Popova, EAMDA Executive Committee, Bulgarian Association for Neuromuscular Diseases & EPF Youth Group, Bulgaria

Inmaculada Placencia Porrero, Senior Expert Social Affairs, European Commission, DG for Employment, Social Affairs and Inclusion, Unit for Disability and Inclusion

Prof. Jerome Bickenbach, Professor Emeritus at Queen's University, Canada and Visiting Professor at the University of Lucerne

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SESSION 0204: Friday 15th May 2020, 14:00 – 15:30

Reaching Future Scenarios: From Accidental Progress to Success by Design

This session will present the scenarios currently being developed under the Rare2030 project and the different policy options associated to them. The session aims to be interactive, engaging the panellists and audience in a back-casting exercise and a vote on the preferred policy options.

Chair: Hans Winberg, Secretary General, Leading Health Care, Sweden

Speakers:

Terkel Andersen, President, EURORDIS-Rare Diseases Europe

Rebecca Tvedt Skarberg, Advisor, Osteogenesis Imperfecta Federation Europe (OIFE), Norway

Prof. Milan Macek, Dept of Biology and Medical Genetics, Charles University and Orphanet Czech Republic

Anna Krohwinkel, Leading Health Care, Sweden

THEME 3

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

THEME LEADERS:

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

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

Matt Bolz-Johnson, ERN and Healthcare Advisor, EURORDIS

THEME SUPPORT :

Ines Hernando, ERN and Healthcare Director, EURORDIS

THEME DESCRIPTION:

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

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

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

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

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

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

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

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

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

Speakers:

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

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

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

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

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

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

ERN & CoE Accreditation as Quality Improvement Framework

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

Identification of experts in each and every Member State (MS) is the first step towards securing an accurate diagnosis and accessing appropriate care. National processes for endorsing rare disease expert

centres continue to be developed across EU MS. Endorsement and accreditation is a dynamic quality improvement process that incrementally raises the quality thresholds services need to meet to be approved. The future trend will be two-fold - universal coverage of national accreditation of highly specialised healthcare and rare diseases centres, and the incremental step-wise maturing of the accreditation process to come to a final result that will be measured on treatment outcome.

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

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

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

Speakers:

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

Anke Widenmann-Grolig, KEKS & EAT, Germany

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

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

Clinic of the Future & Digital Care Pathways

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

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

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

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

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

Speakers:

Melina Brovall, Cystic Fibrosis Patient Representative, Sweden

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

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

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

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

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

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

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

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

For those with rare diseases there cannot be any doubt that greater inter-connectivity and patient power has been beneficial. However there is potentially a fly in the ointment - there's a difference between a

patient who's done a rudimentary google search about some transient symptoms and an expert patient who is one of a handful with a specific condition in the country. How will doctors be able to distinguish between the two when they come to them holding out printed information to be read and understood in a 10-minute appointment? Is the term "expert patient" being devalued?

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

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

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

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

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

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

Speakers:

Prof. James Buchan, Queen Margaret University Edinburgh, UK

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

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

THEME 4

WHEN THERAPIES MEET THE NEEDS: ENABLING A PATIENT-CENTRIC APPROACH TO THERAPEUTIC DEVELOPMENT

THEME LEADERS:

Diego Ardigo, Global Rare Diseases R&D Head, Chiesi Farmaceutici, Italy & Chair, Therapies Scientific Committee of IRDiRC

Virginie Hivert, Therapeutic Development Director, EURORDIS and vice-Chair, Therapies Scientific Committee of IRDiRC

THEME SUPPORT:

Maria Cavaller, Patient Engagement Junior Manager, EURORDIS

THEME DESCRIPTION:

The objectives of this theme are to take stock of the experience gained so far in the development of medicines for people living with rare diseases, and to examine the evolution of the field. We will look at recent scientific innovations and clinical research, regulatory solutions, roadblocks and challenges in developing therapies that match the needs of the patients, as well as ways of embedding real life evidence into the therapeutic development processes.

PRE-RECORDED SESSION: (Available on demand)

Galaxy Guide for Rare Disease therapies development

Presenters: Dr. Diego Ardigo, Global Rare Diseases R&D Head, Chiesi Farmaceutici, Italy and Chair, Therapies Scientific Committee of IRDiRC & **Dr. Virginie Hivert,** Therapeutic Development Director, EURORDIS & Vice-Chair, Therapies Scientific Committee of IRDiRC

The Orphan Drug Development Guidebook Task Force was set up within the International Rare Disease Research Consortium (IRDiRC) with the aim to create a guidebook describing the available tools and initiatives specific to rare disease development and how to best use them, in order to address the multiple challenges inherent to drug development for rare diseases for which the traditional model of development is becoming less and less suitable.

The aim of the Guidebook is to benefit the various stakeholders working in the orphan drug development environment. It includes fact-sheets describing each tool or resource (covering a large number of initiatives that are available worldwide), a series of standard use cases defining how and when to use them, and a series of practical checklists of items to consider at each step of the development. Integration of such elements within a defined drug development framework is set out to generate better data quality, shorter development timelines, and better R&D efficiency.

SESSION 0401: Thursday 14th May 2020, 14:45 – 16:15

What do patients expect from therapy development?

This session aims to look at the present and future of medicine's development and to reflect what patients are expecting for the next decade.

Building on results from the Rare Barometer Surveys (e.g. RD patients' experience with accessibility to treatments), the current work around patient engagement and a few figures illustrating the current state-of-play of therapies development for rare diseases, will also help to set the scene.

A panel of patients representing different rare disease areas, with expertise in several aspects of the medicine's life-cycle and engaging with the ecosystem in a variety of capacities (EMA, EURO-CAB, IMI PARADIGM on sustainable patient engagement, ERNs, HTA, etc.) will discuss the actual challenges, the needs and main expectations vis-à-vis the development of medicines, and the way for each stakeholder to contribute to improving RD patients' lives.

Chair: Dr. Virginie Hivert, Therapeutic Development Director, EURORDIS & Vice-Chair, Therapies Scientific Committee of IRDiRC

Speakers:

Loris Brunetta, Thalassaemia International Federation, Italy

Alain Cornet, Lupus Europe, Belgium

Veronica Popa, MCT8-AHDS Foundation, Greece

Eva Stumpe, SMA Europe, Germany

Russell Wheeler, Leber's Hereditary Optic Neuropathy Society, UK

SESSION 0402: Thursday 14th May 2020, 16:45 – 18:15

Disruptive Innovations in clinical research

This session will focus on innovative trends in clinical research, both in study design and execution, as well as innovative approaches to data collection. We will discuss the opportunities and challenges posed by these developments, together with the challenges foreseen in terms of regulatory and HTA assessment and the impact for the rare disease patients.

Attendees will leave this session with a broader view and understanding of the opportunities and challenges generated by current changes to how clinical research is conceived and executed, and the impact these changes will have on evidence generation in the future.

Chairs: Dr. Diego Ardigo, Global Rare Diseases R&D Head, Chiesi Farmaceutici, Italy and Chair, Therapies Scientific Committee of IRDiRC & **Dr. Violeta Stoyanova-Beninska**, Chair, Committee for Orphan Medicinal Products, EMA

Speakers:

Dr. Simon Day, Clinical Trials Consulting & Training, UK

Dr. Nigel Hughes, Janssen Research and Development, Belgium

Prof. Armando Magrelli, Istituto Superiore di Sanità, Italy

Elizabeth Vroom, World Duchenne Organization, Netherlands

Pooja Merchant, Bayer, USA

Prof. Veronica Miller, University of California, USA

SESSION 0403: Friday 15th May 2020, 11:30 – 13:00

Innovation in Advanced Therapy

In this session, we will dive into the development and use of an ATMP from idea to approval and beyond. Using a particular product as an example, we will look at it from every angle: developer, patient, physician and regulator. Participants should leave this session with a greater understanding of the challenges relating to developing an ATMP and the subsequent use of it in clinical practice, which can be rather different from a standard product.

Chair: Dr. Kristina Larsson, Head of Orphan Medicines, European Medicines Agency

Speakers:

Tomasz Grybek, Fundacja Bohatera Borysa (Boris the Hero Foundation), Poland

Michela Gabaldo, Head of Alliance Management & Regulatory Affairs, Fondazione Telethon, Italy

Patrick Célis, European Medicines Agency, The Netherlands

SESSION 0404: Friday 15th May 2020, 14:00 – 15:30

Bringing real life into therapeutic development

The patient should be central to all aspects of drug discovery, development, regulatory approval and future evidence generation of medicines, ensuring a complete life cycle approach to patient engagement. This session showcases where patients can bring their real-life experiences into different areas of the drug development pathway. We will explore how patient engagement in clinical trial design is increasingly valued, ensuring that studies capture what is important to measure from the end user perspective.

Patient reported outcomes (PRO) measure how a patient feels and functions whilst on a therapy. Developing PRO standards ensures robust data collection and interpretation, adding value to the information available about the patient experience whilst on a therapy.

The European Reference Networks facilitate discussions on rare diseases, concentrating knowledge and allowing for the collection of real world data which can be used to learn more about rare conditions and available therapies. Drug repurposing is a hot topic and an area where rare disease groups are now often leading the way, directing the development pathway for the benefit of their patient group.

Chairs: Dr. Daria Julkowska, Assistant Director, Institute GGB; Coordinator, European Joint Programme on Rare Diseases & **Dr. Daniel O'Connor,** Medical Assessor, Medicines and Healthcare Products Regulatory Agency (MHRA)

Speakers:

Dr. Anja Schiel, Norwegian Medicines Agency

Prof. Faisal Ahmed, Endo-ERN EuRRECa, UK

Dr. Madeline Pe, EORTC, Belgium

Dr. Nick Sireau, AKU Society, UK

THEME 5

ACHIEVING THE TRIPLE AS BY 2030: ACCESSIBLE, AVAILABLE AND AFFORDABLE THERAPIES FOR PEOPLE LIVING WITH A RARE DISEASE

THEME LEADERS:

Dimitrios Athanasiou, European Medicines Agency, World Duchenne Organisation and European Patient Forum, Greece

Prof. Josep Torrent-Farnell, Universitat Autònoma de Barcelona, Spain

THEME SUPPORT:

Simone Bosseli, Public Affairs Director, EURORDIS

Ana Palma, Senior Director Global HTA & Patient Access Lead, SOBI

THEME DESCRIPTION:

There are more life-changing therapies in development for people living with rare diseases than ever before, yet at our current pace it will still take decades to cover all our unmet needs. The rare disease community still faces a number of challenges in accessing authorised therapies, which indicates that the system in its current design is not functioning to the benefit of all, particularly those people living with a rare disease.

How can we improve the functioning of the system by 2030? What are the solutions to ensure the sustainable development of therapies that are truly available to all? This theme will examine the different aspects of the system which need significant change to make it 'fit-for-purpose' now and for our future needs.

SESSION 0501: Thursday 14th May 2020, 14:45 – 16:15

Rare Diseases in Numbers: What do they mean?

There is a growing need for accurate baseline numbers to enable effective evidence-based advocacy for the rare disease community. Recent initiatives have addressed this need for data, with regard to the key issues of access, the economic burden of rare diseases, and the budgetary impact of therapies. This session will discuss recent studies and methodologies related to these issues.

Chair: Avril Daly, CEO, Retina International; Vice-President, EURORDIS

Speakers:

Dr. Ana Rath, Director, Orphanet

Sandra Courbier, Social Research Director - Rare Barometer Programme Lead, EURORDIS-Rare Diseases Europe

Dr. Orla Galvin, Director of Research Policy, Retina International, Dublin

Alexander Natz, Secretary-General of EUCOPE, Belgium

SESSION 0502: Thursday 14th May 2020, 16:45 – 18:15

New disruptive technologies: how can we prepare healthcare systems?

Gene and cell therapies (ATMPs) have the potential to bring a level of disruption to treatment for rare diseases that we have never seen before. This session will explore novel treatments for haemophilia, Spinal muscular atrophy (SMA), thalassemia and retinal disorders, and will feature work done on assessment, availability, access and affordability as part of RARE IMPACT. The panel will discuss their suggestions and potential solutions for improving access across Europe.

Chair: Dr. Mariette Driessens, Policy Officer, VSOP (Dutch Genetic Alliance)

Speakers:

Prof. Hildegard Büning, European Society for Gene and Cell Therapy (ESGCT), Germany

Simone Boselli, Public Affairs Director, (EURORDIS)

Adam Hutchings, Managing Director, Dolon Ltd

Declan Noone, European Haemophilia Consortium, Ireland

Evert Jan Van Lente, AOK Health Insurance, Germany

Lonneke Timmers, Advisor, Zorginstituut Nederland (ZIN), the Netherlands

Francis Pang, Vice President, Global Access, Orchard Therapeutics, UK

SESSION 0503: Friday 15th May 2020, 11:30 – 13:00

From Research to Access: Is a European Collaborative Approach Possible?

Bearing in mind technological advances as well as the need to increase the number of therapies available, can we realistically imagine one seamless European approach from development to access? What elements would this require? Can it be established in the next 10 years? This session will look at the existing successful model of partnership.

Chair: Dimitrios Athanasiou, World Duchenne Organisation

Speakers:

Dr. Elena Nicod, Dolon Ltd, Italy

Toon Digneffe, Head of Public Affairs and Public Policy - Rare Disease Franchise Europe & Canada, Takeda, Belgium

Dr. Donato Bonifazi, Consorzio per Valutazioni Biologiche e Farmacologiche, Italy

Josie Godfrey, Director, JG Zebra Consulting, UK

Dr. Daria Julkowska, Assistant Director, Institute GGB; Coordinator, European Joint Programme on Rare Diseases

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SESSION 0504: Friday 15th May 2020, 14:00 – 15:30

Ensuring Faster Development and Equitable Access: Future Scenarios from Rare 2030

We are seeing emerging narratives emphasising the strain that people living with a rare disease place on the overall healthcare system; yet at the same time, the general public continue to respond in their thousands to crowdfunding appeals, demonstrating an unprecedented sense of solidarity. Crowdfunding is, however, an unsustainable approach. How much is society willing to pay in 2030 for people living with a rare disease? Do we need a solidarity pact? Which future trends in rare disease therapies need to be taken into consideration?

Chair: Sheela Upadhyaya, HST and Topic Selection Specialist Centre for Health Technology Evaluation, National Institute for Health and Care Excellence, UK

Speakers:

Dimitrios Athanasiou, World Duchenne Organisation

Dr. Mariette Driessens, Policy Officer, VSOP (Dutch Genetic Alliance)

Avril Daly, CEO, Retina International; Vice-President, EURORDIS

Giovanna Giuffrè, Project Manager, ISINNOVA, Italy

Ana Palma, Senior Director Global HTA & Patient Access Lead, Swedish Orphan Biovitrum BVBA/SPRL, Belgium

THEME 6

THE DIGITAL HEALTH REVOLUTION: HYPE VS. REALITY

THEME LEADERS:

Julián Isla, Data and Artificial Intelligence Resource Manager, Microsoft & Chief Scientific Officer, Dravet Syndrome European Foundation, Spain.

Justina Januševičienė, Head of Healthcare Innovation Development Centre, Lithuanian University of Health Sciences, Lithuania.

Brian O'Connor, Chair, European Connected Health Alliance, UK/Ireland

THEME SUPPORT :

Denis Costello, Executive Director, CML Advocates Network, Spain.

Marta Campabadal, RareConnect Manager, EURORDIS

THEME DESCRIPTION:

This theme examines the technological innovations that are underpinning disruption in medicine and science, as well as the legal, ethical and policy foundations that can frame future outcomes in this area. The theme will also look at how technology can support the social inclusion of people living with a rare disease.

Attendees should come away from this theme with a greater understanding of the role of quality data in technologies such as Artificial Intelligence and how this impacts the development of medicines and delivery of care and other services. The theme aims to question the value of such technological innovations, as well as to show the policy frameworks and ecosystems which patient representatives can involve themselves in, in order to bring the patient's voice to the evolution of policy and ethics in this area.

SESSION 0601: Thursday 14th May 2020, 14:45 – 16:15

Do we need a New Patient Data Management Model for AI?

Artificial Intelligence seems to be the holy grail, promising to solve the many challenges faced by the current healthcare system. What do we need in order for models based on Artificial Intelligence to work? The answer: large datasets for training those models. These large datasets have usually been safeguarded by hospitals, the traditional centres of medical care.

But this centralized hospital data model is giving way to a hybrid system, where data is generated not only by the hospital but also by the patient and other sources. In addition we have the problem that certain data (such as genomic data) is too large to be stored within the traditional medical information system based in hospitals. Perhaps we need new data management models that allow the capture, processing and analysis of medical data generated by various sources. This question is particularly significant for rare disease patients as they often have to visit multiple specialists in different healthcare centres. In this session we will discuss the new data management models we will need, and potential challenges that may arise (medical, regulatory, security, privacy, ethics).

Chairs: Denis Costello, Executive Director, CML Advocates Network, Spain

Speakers:

Paul Rieger, Managing Director, Centiva Health, Austria

Peter Speyer, Head of Products, data42, Novartis, Switzerland

Dr. Marco Roos, GO FAIR, Netherlands

Nicholas Becker, AI for Good, Microsoft, USA

SESSION 0602: Thursday 14th May 2020, 16:45 – 18:15

Considerations Surrounding the Ethical and Legal Aspects of Data Governance

The current COVID-19 crisis has brought into sharp relief some of the big challenges on the ethical and legal aspects of data governance, most significantly in the debate on the use contact tracing apps as a key tool in the transition out of lockdown and the management of a 'new normal'. The use of such apps highlights the fragile and complex balance between public and private interests in data, the need to build trust in data governance systems and the need for effective mechanisms for data solidarity. In this session you will hear speakers address all of these issues, providing insights on how the balance between public and private is seen in our rare disease communities as well as about exciting developments in new approaches to making data available for research (Findata) and facilitating data solidarity (Salus Coop).

Chair: Dr. Petra Wilson, Managing Director and CEO, Health Connect Partners, UK

Speakers:

Dr. Bettina Ryll, Founder and President, Melanoma Patient Network Europe

Rosa Juuti, Senior Specialist, Findata, Finland

Sandra Courbier, Social Research Director, Rare Barometer Voices, France

Joan Guanyabens, Consultant Health IT and Innovation, Salus Coop, Spain

SESSION 0603: Friday 15th May 2020, 11:30 – 13:00

Getting Solutions to Patients Quicker and more Effectively: Are Policy, Procurement and Patients the Enablers or the Obstacles?

What changes are necessary to get solutions to patients quicker and more effectively? The speakers will share their own experiences and express their own perspectives on whether or not policy, procurement and patients are the enablers or obstacles. This interactive session will seek consensus on concrete actions to overcome any obstacles and map out a way forward.

Chair: Brian O'Connor, Chair, European Connected Health Alliance, UK/Ireland

Speakers:

Prof. George Crooks, Digital Health & Care Institute, Scotland

Jaana Sinipuro, Project Director, IHAN – Human-Driven Data Economy, SITRA, Finland

Liz Ashall-Payne, Founder and CEO, ORCHA, UK

SESSION 0604: Friday 15th May 2020, 14:00 – 15:30

Technology for inclusion and empowerment

This session will highlight the possibilities of empowerment and inclusion presented by innovative technology. You will hear about a number of good examples around tech-tools, digital accessibility and policymaking that can be used as means and stepping stones to reach full holistic inclusion.

Chairs: Robert Hejdenberg, President, Agrenska

Speakers:

Dr. Stefan Johansson, KTH Royal Institute of Technology, Sweden

Daniel Forslund, Assistant Regional Council in the Stockholm Region, Sweden

Allison Watson, Patient Advocate, Ring20, UK

Veronica Popa, Patient Advocate, Romania

Ana Neacşu, Speed (Speech & Dialogue Research Laboratory), Romania

Tim Buckinx, Founder and CEO, Epihunter, Belgium

Michael Lovgren, CEO Assistant, Agrenska, Sweden

Fredrik Ruben, Chief Executive Offficer, Tobii Dynavox, Sweden

CLOSING PLENARY SESSION AGENDA

Friday, 15 May 2020 - 16:00 - 18:00

MODERATOR

Lene Jensen, Director, Rare Diseases Denmark

HIGHLIGHTS AND TAKE-HOME MESSAGES FROM THE PARALLEL THEMES:

- **Theme 1: Prof. Christine Patch,** Clinical Lead for Genetic Counselling, Genomics England

- **Theme 2: Maria Montefusco,** President, Rare Diseases Sweden

- **Theme 3: Dr. Birute Tumiene,** Clinical Geneticists & Coordinator for Competence Centres, Vilnius University Hospital Santaros Clinics, Lithuania

- **Theme 4: Dr. Diego Ardigò,** Chair, Therapies Scientific Committee of IRDiRC and Global Rare Disease R&D Head, Chiesi Farmaceutici, Italy

- **Theme 5: Ana Palma,** Senior Director Global HTA & Patient Access Lead, Swedish Orphan Biovitrum BVBA/SPRL, Belgium

- **Theme 6: Denis Costello,** Executive Director, CML Advocates Network, Spain

PERSONAL TAKE-HOME MESSAGES

Jana Popova, Young Patient Advocate, Bulgarian Association for Neuromuscular Diseases; European Patients' Forum Youth Group; European Alliance of Neuromuscular Disorders

POLITCAL SPEECH

Adam Vojtěch, Minister of Health, Czech Republic

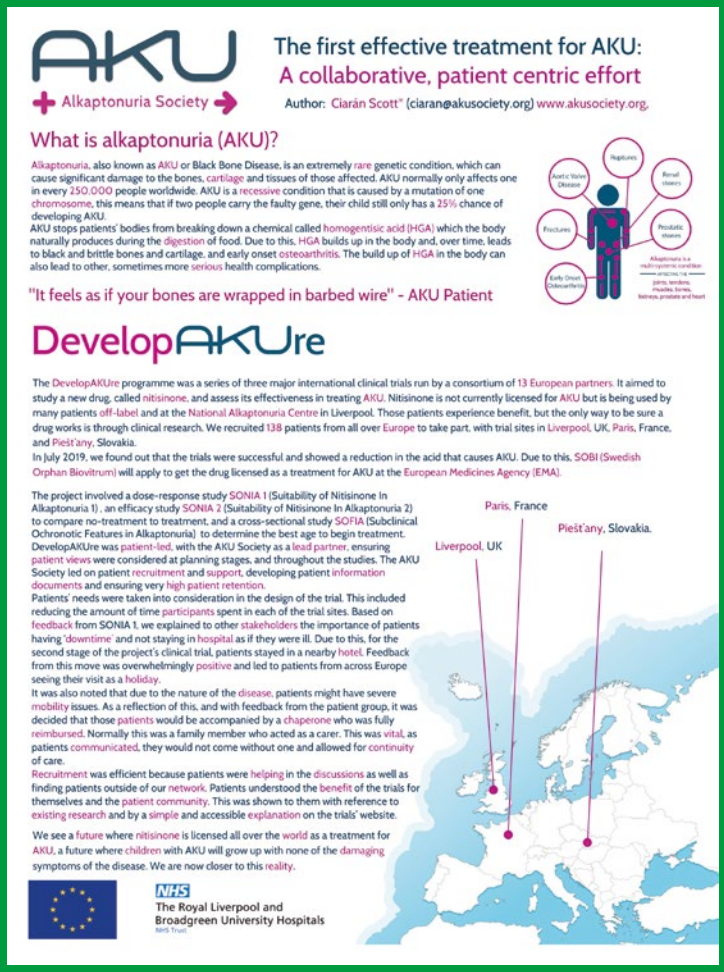
CONCLUSIONS

'Policy Options Emerging from ECRD 2020 on the Journey of Living with a Rare Disease in 2030'

Yann Le Cam, Chief Executive Officer, EURORDIS

POSTERS

Poster Award Presentation; Friday 15 May 10:00 – 11:00 CEST

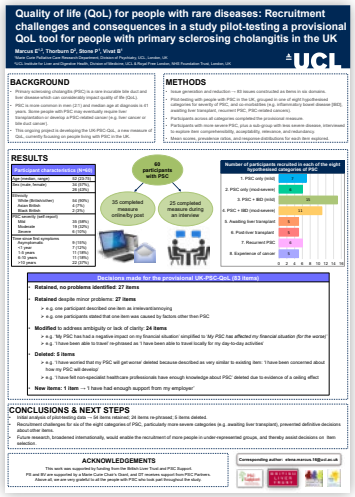


1st Place: P 163

A collaborative and patient-centric effort to find the first effective treatment for alkaptonuria. (Nick Sireau)

2nd Place: P 267

'Patient Journeys': Personal experiences shaping clinical priorities (Olivia Spivack)



3rd Place: P 268

Quality of life (QoL) for people with rare diseases: Recruitment challenges and consequences in a study pilot-testing the UK-PSC-QoL, a provisional QoL tool for people with primary sclerosing cholangitis (PSC) in the UK (Elena Marcus)

POSTERS

THEME 1: THE FUTURE OF DIAGNOSIS: NEW HOPES, PROMISES AND CHALLENGES

P1 | Bayer Pharmaceutical
Genomic testing in the emerging era of precision medicine: Lessons learned from studies in larotrectinib
Jo Ballot
Lauren Kaplanis, Suzanne Ezrre, Tatiana Norman-Brivet, Genevieve Kelly, Sandra Montez

P2 | Mendelian
Case report: Reducing the diagnostic odyssey in Behcet's disease through a digital health approach into primary care UK
Orlando Buendia
Rudy Benfredj, Timothy Halford, Peter Fish, Will Evans, Renate Apse, Isabella Rustignoli

P3 | Mendelian
Case report: cost-benefit of earlier diagnosis in a patient with Behcet's disease using CTV3 codes at primary care UK
Orlando Buendia
Rudy Benfredj, Tim Halford, Peter Fish, Will Evans, Nicolas Aussel, Renate Apse, Isabella Rustignoli

P4 | National Alliance for Rare Diseases Romania
Rare genetic diseases in Timis Regional Centre of Medical Genetics, Romania
Maria Puiu
Adela Chirita Emandi, Iulia Jurca Simina, Alexandra Mihailescu, Nicoleta Andreescu, Dorica Dan

P8 | Dep.of Pediatric Nephrology, RoMed Kliniken, Rosenheim
Molecular-Based Newborn Screening for Cystinosis and Spinal Muscular Atrophy (SMA) in Germany
Hohenfellner,Katharina
Hohenfellner K., Burggraf S., Vill K., Bergmann C., Fleige T., Janzen N., Czibere L., Froschauer S., Röschinger W., Olgemöller B., Nennstiel U.

P10 | asociacion internacional de familiares y afectados de lipodistrofias AELIP
«The Importance of Diagnosis in Lipodystrophies»
Jose Jerez Ruiz
Juan Carrion Tudela

P11 | Medical Informatics Group, University Hospital Frankfurt
Visualization techniques in a Clinical Decision Support System for Rare Diseases
Jannik Schaaf
Martin Sedlmayr, Martin Boeker, Hans-Ulrich Prokosch, Holger Storf

P12 | Karolinska Institutet
Siglec-6: a potential new biomarker for clonal mast cell diseases
Andrea Renate Teufelberger
Andrea R. Teufelberger, Catherine Overed-Sayer, Jingya Wang, Maria Ekoff, Barbro Dahlén, Theo Gülen, Gunnar P. Nilsson

P15 | Centre of rare diseases southeast region
Mind the gap - Rare transition from child to adult care
Åsa Lundin
Charlotte Lilja

P16 | Fondazione Telethon
We made the therapies, now give us newborn screening!
Annamaria Zaccheddu
Alessandra Camerini, Ermanno Rizzi

P18 | Karolinska Institutet
Molecular studies of intestinal malrotation
Karin Salehi Karlslätt
Anna Lindstrand, Britt Husberg, Tomas Wester, Agneta Nordenskjöld

P19 | Medical university Vienna
Volumetry of the anal sphincter complex in infants with anorectal malformation
Patricia Feil
Fartacek, Metzelder, Weber, Patsch, Krois

P20 | Portuguese Association for Congenital Disorders of Glycosylation (APCDG), CDG&Allies-PPAIN
CDG Diagnosis: A simplified guide for different stakeholders
Carlota Pascoal
Dorinda Marques-da-Silva, Rita Francisco, Sandra Brasil, Vanessa dos Reis Ferreira, Ea Morava, Jaak Jaeken

P21 | Service of Genetics Laboratory, Faculty of Medicine, University Hospital center»Mother Teresa', Tirana, Albania
A microdeletion syndrome at 9q21.11-q21.31 characterised by mental retardation, speech delay, epilepsy.
Anila Laku

P22 | Kindernetzwerk e.V.
Autism, Epilepsy and Genetics - An experience with the gene SCN2A
Nicolas Lorente

P23 | Bambino Gesù Children's Hospital in Rome
Timeliness of Genetic Diagnosis in Critical Newborns: Proposal of a Dedicated Diagnostic Path
Cortellessa Fabiana
Digilio Maria Cristina, Dallapiccola Bruno, Novelli Antonio, Agolini Emanuele, Lepri Francesca, Dotta Andrea, Bagolan Pietro, Macchiaiolo Marina, Bartuli Andrea.

P24 | CIBERER (Centre for Biomedical Network Research on Rare Diseases)
The CIBERER Program for Undiagnosed Rare Diseases (ENOD). A collaborative and multidisciplinary approach to diagnosis
Beatriz Morte
Morte B., Moreno E., Herreras E., Pérez-Florido J., Dopazo J, Pérez-Jurado L.A.

P25 | Karolinska Institutet
Germline mutation c.4031CT (S1344L) in the RNase IIIa domain of DICER1 causes GLOW syndrome
Emeli Pontén
Emeli Pontén, Sofia Frisk, Kristina Lagerstedt-Robinson, Ann Nordgren

P28 | GIIS-012. Instituto de Investigación Sanitaria Aragón (IIS Aragón), Unidad de Investigación Traslacional.
Next Generation Sequencing (NGS) an opportunity for patients non-diagnostic in Lysosomal Storage Disease
Isidro Arévalo-Vargas
López de Frutos, Serrano-Gonzalo, M Strunk, P Mozas, JJ Cebolla, P Giraldo.

P29 | Karolinska Institutet
Genetic diagnoses in skeletal ciliopathies using MPS, structural variant, and RNA analyses
Dominyka Batkovskyte
Anna Hammarsjö, Maria Pettersson, David Chitayat, Atsuhiko Handa, Fulya Taylan, Dominyka Batkovskyte, Britt-Marie Anderlid, Angela E. Lin, Kenji Shimizu, Ana Beleza-Meireles, Brian Hon-Yin Chung, Ulrika Voss, Ann Nordgren, Gen Nishimura, Anna Lindstrand, Giedre Grigelioniene

P30 | Ågrenska AB
Stiftelsen Sällsyntafonden - The Rare Disease Research Foundation
Ann Nordgren
Anders Olauson, Robert Hejdenberg and Maria Montefusco

P31 | EURORDIS-Rare Diseases Europe
Mapping of Resources from Networks for Undiagnosed and Newly Diagnosed Ultra-Rare Diseases
Gulcin Gumus
Gulcin Gumus, Stephanie Broley, Christine Patch, Marisol Montolio del Olmo, Kym Boycott, Laurence Faivre, Birte Zurek, Milan Macek, Holm Graessner, Simona Bellagambi, Daphne Stemkens, Cathalijne van Doorne, Alison Metcalfe, Alessia Costa, Glenn Robert, Lauren Roberts, Marie Pritchard, Hans Scheffer, Vanessa Boulanger, Janine Lewis, Helen Cederroth, Mikk Cederroth, Patricia Arias, Virginie Bros Facer

P32 | AveXis, Inc.
Cost-Effectiveness Analysis of Newborn Screening for Spinal Muscular Atrophy (SMA) in the United States
Anish Patel
Ramesh Arjunji, Jenny Zhou, Marie Louise Edwards, Michael Harvey, Eric Wu, Omar Dabbous

POSTERS

P33 | Karolinska Institutet
A case of ring chromosome 21 with hemifacial microsomia, developmental and speech delay analysed with WGS and patient-derived NES cells
Jakob Schuy
Jesper Eisfeldt, Maria Pettersson, Niloofar Shahrokhshahi, Mansoureh Shahsavani, Daniel Nilsson, Anna Falk, Anna Lindstrand

P34 | Karolinska Institutet
Whole genome sequencing discovers clonal evolution of leukemic clones from birth to the time of diagnosis of concordant acute lymphoblastic leukemia in a monozygotic twin pair
Fulya Taylan
Fulya Taylan, Benedicte Bang, Arja Harila-Saari, Jesper Eisfeldt, Mats Heyman, Gisela Barbany, Vasilios Zachariadis, Ann Nordgren

P35 | Sciensano
Genetics in Belgium: Today And Tomorrow
Joséphine Lantoine
Jean-Bernard Beaudry and Nathalie M. Vandevelde

P36 | AISMME Associazone Italiana Sostegno Malattie Metaboliche Ereditarie APS (Italian Association Inherited Metablic Diseases Support)
Extended newborn screening
MANUELA VACCAROTTO
Giuliana Valerio

P37 | Wales Gene Park
Engaging and Involving the Rare Disease community in Wales through Genomics Cafes
Rhian R Morgan
Emma L Hughes, Angela M Burgess

P38 | Genetic Alliance UK/ Wales Gene Park
Co-production of the Welsh Rare Disease Research Gateway
Emma L Hughes
Rhian R Morgan, Angela M Burgess

P39 | Genetic Alliance UK
Genomic data and the NHS: views of rare disease patients and carers
Amy Hunter
Rosa Spencer-Tansley, Simon Wilde

P40 | FEDER – Federación Española de Enfermedades Raras
The reality of undiagnosed 'Rare Patients' - claiming for an improvement in their life-quality
Alba Ancochea

P41 | UNIAMO FIMR
Position Paper on Extended Newborn Screening by the Italian Associations of People Living with Rare Diseases
Annalisa Scopinaro

P42 | Karolinska Institutet
KIAA0753 mutations in skeletal ciliopathies: unveiling disease mechanisms
Raquel Vaz
Anna Hammarsjö, Fulya Taylan, David Chitayat, Giedre Grigelioniene, Anna Lindstrand

P43 | Monash University Malaysia
A Novel Globotriaosylceramide Quantification Assay for the Rapid Diagnosis of Fabry Disease
Allan Ng Wee Ren
Kumaran Narayanan

P277 | Swiss Society for Porphyria
Abdominal Pain «Without a Good Reason» - Think Acute Hepatic Porphyria!
Jasmin Barman-Aksözen, PhD, 1,2
Eva Schupp, Marten Pettersson, Mehmet Aksözen PhD, Francesca Granata PhD, Cornelia Dechant MD, Rocco Falchetto PhD

THEME 2: OUR VALUES, OUR RIGHTS, OUR FUTURE: SHIFTING PARADIGMS TOWARDS INCLUSION

P45 | The Finnish Network for Rare Diseases
Advancing meaningful patient involvement: case HARKKO patient advocacy group
Carita Åkerblom, Matti Santalahti
Carita Åkerblom, Members of Harkko patient advocacy group

P46 | Cystic Fibrosis Ireland
"I may have CF, but it does not have me" Independent living and Cystic Fibrosis
Sarah Tecklenborg
Rory Tallon, Gerry Walker, Fiona Bodels, Rebecca Horgan, Samantha Byrne, Caroline Heffernan, Daniel Costigan, Philip Watt

P47 | University College Dublin
Children's voices in Rare Diseases - Sand play, arts, music, photo voice and interviews
Dr Sandra Mc Nulty
Somanadhan, S., McNulty, S., Karpaviciute, S., O'Neill, A., Sweeney, A. Brinkley, A., O'Toole, D., Bracken, S., Flynn, C., Larkin, P., Awan, A., Baker. M, Kroll, T.

P48 | RVRH-XLH, French association of patients with XLH
Unmet needs of adults with XLH: results from a survey of European patient organizations
Pol Harvengt
Lothar Seefried, Lindsay Perera

P50 | Ersta Skondal Bracke University College
Views on everyday life among adults with spina bifida: an exploration through photovoice
Hanna Gabrielsson
Eric Asaba, Agneta Cronqvist

P51 | University College Dublin
Measuring parenting stress, quality of life and resilience related to caring for children, young people and adults living with Mucopolysaccharidosis (MPS)
Suja Somanadhan
Bristow, H., Brinkley, A., Crushell, E., Larkin, P., Nicholson, E., Pastores, G., Somanadhan, S1.

P52 | Vifor Fresenius Medical Care Renal Pharma Ltd.
SEE ME. HEAR ME: Support for anyone living with, or affected by, ANCA-associated vasculitis (AAV)
Peter Verhoeven
Julie Power, John Mills (MD), Dijana Krafcsik, Peter Rutherford (MD), Dieter Götte (MD)

P53 | IRCCS Istituto Ortopedico Rizzoli
TemeRARI si nasce - Born to be Brave
Manila Boarini
Manila Boarini, Andrea Romeo, Silvio Boero, Davide Scognamiglio, Luca Sangiorgi, Eleonora Grippa

P54 | Bambino Gesù Children's Hospital (Rome -Italy)
Empowerment Intervention for persons affected by Williams Syndrome: a pilot project at the Bambino Gesù Children's Hospital
Francesca De Lorenzo
De Lorenzo F., Macchiaiolo, M., Buonuomo, P.S., Capolino, R., Dentici, M.L., Digilio M.C., Rana, I., Bartuli, A.

P55 | International Prader-Willi Syndrome Organisation (IPWSO)
Addressing the Ethical and Therapeutic Challenges of a Rare Disability: Hyperphagia and Other Behaviours in People with Prader-Willi Syndrome
Anthony Holland
Elizabeth Fistein, James O'Brien, Maria Libura

POSTERS

THEME 3: SHARE, CARE, RARE: TRANSFORMING CARE FOR RARE DISEASES BY 2030

P56 | Vilnius University Hospital Santaros Klinikos
Inpatient Day Centre in Pediatric Unit of Vilnius University Hospital Santaros Klinikos
Viktoras Sutkus
Dovile Kalibatiene, Paule Stulginskaite, Rimante Cerkauskiene

P57 | Public Health England
Population based data collection of Wilson's Disease in England through a national rare disease register
Osob Mohamed
Mary Bythell, Jeanette Aston, Sarah Stevens

P58 | Wroclaw Medical University, Department of Pediatrics
Health Related Quality Of Life After Surgical Repair Of Esophageal Atresia
Anna Rozensztrauch
Anna Rozensztrauch, Robert Smigiel, Dariusz Patkowski

P59 | CDH International
25 Year Retrospective on Congenital Diaphragmatic Hernia
Dawn Ireland
Tracy Meats, Kamal Salah, Darlene Silverman

P60 | The Office for Rare Conditions
The use of patient reported experiences to develop local solutions for individuals with rare and low prevalence conditions in the West of Scotland.
Martina Rodie
Martina Rodie, Elizabeth Dougan, Shannon Mullen, Daisy Johnston, Moncia Hytiris, Fasial Ahmed.

P61 | BridgeBio
Patient and family experience of ATTR Amyloidosis: Results of two focus groups
David Rintell, Ed.D.
Florencia Braga Menéndez, Dena Heath, Jocelyn Ashford.

P62 | Public Health England
NCARDRS: A population-based congenital anomaly and rare disease register for England
Mary Bythell
Jeanette Aston, Jennifer Broughan, Sarah Stevens

P63 | Institut de Myologie, Paris
Screening for neuromuscular diseases: Patient organisations' achievements in European Reference Networks
Dr. Teresinha Evangelista

P64 | European Medicines Agency
Gene Therapy Medicinal Products (GTMPs) represent a paradigm shift in health care as they have great potential for preventing and treating rare diseases with high unmet medical need.
Francesca Tomeo
Segundo Mariz, Armando Magrelli, Violeta Stoyanova

P65 | Salivary Gland Cancer UK
Meeting unmet needs for Adenoid Cystic Carcinoma (ACC) research in the UK and beyond
Emma Kinloch
Robert Metcalf

P66 | European Reference Network RARE-LIVER
Quality of life in patients with autoimmune and cholestatic liver diseases: ongoing agenda in ERN RARE-LIVER NETWORK
Maciej K. Janik
M.K.Janik, E.Wunsch, T.Gevers, J. Willemse, M.Krawczyk, D.Thorburn, Ch.Schramm, A. Lohse, P.Milkiewicz

P67 | Dep. of Pediatric Nephrology, RoMed Kliniken, Rosenheim
Interdisciplinary Cystinosis Clinic Rosenheim – a treatment model for rare multiorgan diseases
Ulrike Treikauskas
Bechtold Dalla Pozza S., Buss M., Dosch R., Erler J., Froschauer S., Goebel J., Herzig N., Holla H., Getzinger T., Knerr C., Koeppl C., Ockert C., Passow M., Priglinger C., Rohayem J., Steidle G., Storf H., Thiele A., Vasseur J., Weber R., Weitzel D., Vill K., Hohenfellner K., and the Germann Cystinosis patient support group

P68 | Institut de Myologie, Paris
A registries hub against data siloing (ERN Euro-NMD Registry)
Dr. Teresinha Evangelista
Teresinha Evangelista, François Lamy, Florence Favrel-Feuillade, Dimitrios Athanasiou, Elizabeth Vroom, Adrian Tassoni, Peter-Bram t'Hoen

P69 | Department of Histology, University "Magna Græcia", Catanzaro, Italy
A novel NIR spectroscopic qualitative method to monitor the diet compliance in PKU patients
Marco Vismara
G. Bonapace, O. Marasco, G. Scozzafava, M. Ashour, M. Pittelli, T. Greto, M.T. Moricca, S.A. Vismara, A. Valentini, D. Concolino, N. Perrotti

P70 | ORPHANET-INSERM
Codification for Rare diseases: the RD-CODE project support to Member States and beyond
Sylvie Maiella
Waed Abdel-Khalek-Haidar, Céline Angin, Virginia Corrochano, Ines Hernando, KurtKirch, Debby Lambert, Monica Mazzucato, Paola Facchin, Stefanie Weber, Miroslav Zvolsky and Ana Rath

P71 | Medical Informatics Group, University Hospital Frankfurt
The ERN-Lung Registry Data Warehouse in the European Reference Network Respiratory Diseases
Holger Storf
Jannik Schaaf, James Chalmers, Heymut Omran, Oliver Sitbon, Martin Dugas, Sarah Riepenhausen, Thomas O.F. Wagner

P72 | Filière de Santé Maladies Rares des Malformations de la tête, du cou et des dents (TETECOU)
The French Rare Diseases Network TETECOU: improving diagnosis and healthcare, promoting education and supporting research for head, neck and tooth malformations
Myriam de Chalendar
Myriam de Chalendar, Inès Ben Aissa, Marie Daniel, Martin Fidalgo, Sandrine Mendy, Marie-Paule Vazquez and Nicolas Leboulanger

P73 | NF Kinder
NF Kinder Center of expertise
Claas Röhl
Amedeo Azizi, Thomas Pletschko

P74 | Universitätsklinikum Hamburg-Eppendorf
Interface management concepts in the health care for rare diseases in Germany: A mixed-methods study to develop best practice recommendations
David Zybarth
Maja Brandt, Ramona Otto, Laura Inhestern, Martin Härter and Corinna Bergelt

P76 | Frambu Resource Centre for Rare Disorders
Health education competence and rare disorders - an online and collaborative academic course on bachelor-level
Christoffer Hals
Gro Trae

P77 | University Hospital Tübingen, Institute of Medical Genetics
Knowledge travels, not trainees and trainers: joint webinar series of EAN, ERN EuroNMD and ERN-RND
Sanja Hermanns
Annemarie Post, Alicia Brunelle, Holm Graessner

P78 | Sahlgrenska University Hospital
National Program Area - Rare diseases: Promoting and enabling knowledge-based, equal and resource-efficient healthcare for rare diseases in Sweden
Anna Wedell
Lovisa Lovmar, Marie Stenmark-Askmalm, Cecilia Gunnarsson, Maria Johansson Soller, Cecilia Soussi Zander, Magnus Burstedt, Anna Zucco, Anna Wedell

P79 | ERN eUROGEN
eUROGEN Share, care, cure. ERN for urorectogenital diseases and conditions
Michelle Battye
Naside Mangir, Darren Shilhan, Jen Tidman, Wout Feitz

P80 | ERN-EuroBloodNet
ERN-EuroBloodNet- Establishment of the European Network of Sickle Cell Disease Patients Organizations
Mariangela Pellegrini
Mariangela Pellegrini, Pierre Fenaux, Béatrice Gulbis, Victoria Gutiérrez Valle, María del Mar Mañú Pereira

POSTERS

P81 | ERN-EuroBloodNet
ENROL, the European Rare Blood Disorders Platform
Victoria Gutiérrez Valle
Victoria Gutiérrez Valle, Béatrice Gulbis, Pierre Fenaux, Mariangela Pellegrini, Marina Kleanthous, Petros Kountouris, Stella Tamana, María del Mar Mañú Pereira

P82 | Heidelberg University Hospital / ERKNet
ERKNET, The European Rare Kidney Disease Reference Network
Vera Cornelius
Tanja Wlodkowski, Giulia.Bassanese, Claudia Sproedt, Franz Schaefer

P83 | UNESCO Chair, School of Health and Social Sciences, Institute of Technology Tralee, Ireland
Living Well with Phenylketonuria (PKU) – the Irish PKU Community Perspective
Bernadette Gilroy
Dr Suja Somanadhan

P84 | HUS
An e-Learning program dedicated to Rare Eye Diseases in Europe, an ERN-EYE initiative
Leroux, Dorothee
Leroux, D., Dollfus, H.

P85 | The Ehlers-Danlos Society
EDS ECHO: Worldwide, Case-based Learning And Support For Clinicians Managing Ehlers-Danlos Syndromes.
Dr Alan Hakim
Dr Clair Francomano, Lara Bloom, Stacey Simmonds, Angela Ballard, Dr Jane Schubart, Dr Rebecca Bascom

P87 | Hope for Hypothalamic Hamartomas-UK; Hope for Hypothalamic Hamartomas
Centralised Efforts Transforming Global Care: The Ten-Year Impact of a Patient Advocacy Group - Medical Expert Partnership on Recognising and Treating a Rare and Complex Epileptic Brain Malformation Syndrome
Emma Nott
Lisa Soeby, Erica Webster, Kathryn Jensen, Kimberly Ranson

P88 | ePAG EpiCARE/Hope for Hypothalamic Hamartomas-UK
The Rare and Complex Epilepsies – Common Unmet Needs within the Patient Community
Allison Watson
Emma Nott, Isabella Brambilla, Torie Robinson, Rosaria Vavassori, Monica Lucente, Barbara Nicol, Carol-Anne Partridge, Anita Noordhoff

P90 | Genetic Alliance UK
How are patients with rare diseases and their carers impacted by the way care is coordinated in the UK? An exploratory qualitative interview study
Amy Simpson
Amy Simpson, Naomi Fulop, Emma Hudson, Stephen Morris, Angus Ramsay, Holly Walton and Amy Hunter.

P92 | University College Dublin
Identifying Research Priorities - Rare Disease Research Partnership (RAinDRoP)
Dr Suja Somanadhan
Somanadhan, S., Nicholson, E., Dorris, E., Brinkley, A., Daly, A., Keenan, A., Awan, A., Ennis, S., Mc Garth, V., Mitchell, D., O'Sullivan, G., Power, J., Treacy, E., Lawlor, A., Harkin, P., Lynch, S.A., Donnelly, S.,Kroll, T.

P93 | The Health Policy Partnership
Expanding the availability of radioligand therapy to meet the treatment needs of people with neuroendocrine neoplasms.
Christine Merkel
Catherine H. Whicher, Nikie Jervis, Ken Herrmann, Jaroslaw Ćwikła, Jamshed Bomanji, Marianne Pavel, Suzanne Wait

P94 | The global Aortic Disease Awareness Day
A Patient Perspective on Aortic Disease
Timo Söderlund
11 people helped in making the paper that is the basis of the poster.

P96 | AveXis, Inc.
Economic Burden of Care and Treatment Options for Patients with Rett Syndrome: Two Systematic Literature Reviews
Omar Dabbous
Omar Dabbous, Vanessa Taieb, Emna Abdennadher, Meryem Bouchemi, Justyna Chorąży, Katarzyna Borkowska, Veneta Georgieva, Bryan E. McGill, Thomas A. Macek, Benit Maru

P97 | Department of Public Health and Caring Sciences, Uppsala University, Sweden
"Nobody could understand what was wrong with me"
Kerstin Hamberg Levedahl
Annika Nilsson, Birgitta Johansson, Mariann Hedström

P98 | Hospital Vall d'Hebron
The JOURNEY of living with a RARE DISEASE in 2030 MetabERN: how we share, how we care
Maurizio Scarpa
Coordinator on behalf of all the MetabERN Members

P100 | Berlin Institute of Health
The Collaboration on Rare Diseases (CORD-MI): A National Initiative to Improve the Documentation and Joint Use of Rare Disease Data in German University Hospitals
Josef Schepers

P101 | University College London
What ways of coordinating care for rare conditions currently exist and are possible? Exploring models of care coordination and stakeholder preferences
Holly Walton
Amy Simpson, Emma Hudson, Angus Ramsay, Amy Hunter, Naomi Fulop, and Stephen Morris.

P102 | HHT Sverige on behalf of HHT Europe
Bridging the Expertise Gap in HHT Emergency Care
Mildred Lundgren
Anne Goustille, Bienvenido Muñoz, Fernando Brocca, Dara Woods, Ralf Schmiedel, Claudia Crocione

P103 | Aix-Marseille Université, INRA, INSERM, C2VN, Marseille, France
Impairment of Vitamin E intestinal secretion in primary hypobetalipoproteinemias: mechanistics studies in a cell model
Claire Bordat
Yan Xie, Nicholas Davidson, Marion Nowicki, Charlotte Halimi, Noël Peretti, Emmanuelle Reboul

P104 | PTC Therapeutics International Limited
Aromatic-L-amino acid decarboxylase (AADC) deficiency: What is it and how is it diagnosed?
Sian O´Niell
Christian Werner MD., Serene Forte PhD, MPH., Axel Boehnke

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Lovisa Lovmar
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P108 | Radboud university medical center
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Matt Bolz-Johnson
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P109 | Radboud university medical center
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P110 | federg- erknet
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Susana Carvajal
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P113 | University Clinic Tuebingen
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Carola Reinhard
Rebecca Schuele-Freyer, Caterina Mariotti, Enrico Bertini, Alfons Macaya, Odile Boespflug-Tanguy, Samuel Groeschel, Ingeborg Krägeloh-Mann, Nicole Wolf, Rik Vandenberghe, Isabelle Le Ber, Markus Otto, Tobias Baeumer, Belén Pérez Dueñas, Giovanna Zorzi, Thomas Gasser, Wassilios Meissner, Anne-Catherine Bachoud-Lévi, Bernhard Landwehrmeyer, Juan Dario Ortigoza Escobar, Ludger Schöls, Holm Graessner

P116 | International Gaucher Alliance (IGA)
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Maddie Collin; Elin Haf Davies

P118 | ERN GENTURIS (ERN on Genetic Tumour Risk Syndromes) & HCP Members (ErasmusMC-Sophia, Rotterdam)
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P119 | ERN ITHACA, EURORDIS, RARE DISEASES Romania & International Federation for Spina Bifida
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Matt Johnson, Yvonne Milne, Ammi Sundqvist, Gabor Pogany, Sue Routledge, Carole Herman

P120 | Hospital Clínic, Barcelona University
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Miquel Blasco
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P121 | CMTC-OVM
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Lex van der Heijden

P122 | Naevus Global
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Yukiko Nishimura
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P125 | Pulse Infoframe Inc.
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Femida Gwadry-Sridhar
C. Elizabeth McCarron, Femida Gwadry-Sridhar

P127 | NPO ASrid
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Shun EMOTO
Kota Ninomiya, Yukiko Nishimura

P128 | European Medicines Agency
Orphan medicinal products for the treatment of pancreatic cancer
Jorn Mulder
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P129 | Kharkiv Charitable Foundation "Children with Spinal Muscular Atrophy", CSMA, Ukraine
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Vitaliy Matyushenko
Andriy Shatillo

P130 | MediTech Media
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Fani Petridis
Senam Beckley-Kartey, Dylan Trundell, Hannah Staunton, Rakesh Kantaria, Florian Lipsmeier, Michael Lindemann, Kristina Gelblin, Nicole Gusset, Mencia de Lemus, Vitaliy Matyushenko, Michael Ostland

P131 | Novartis
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Federica Castiglione
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P132 | LUMC, dept. epidemiology
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Cees SMIT
Cees Smit and Mariëtte Driessens

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Cláudia Hirawat

P134 | Portuguese Association for Congenital Disorders of Glycosylation (APCDG), CDG&Allies-PPAIN
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Carlota Pascoal
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Celia Marín, Jan Geissler, Zack Pemberton-Whiteley, Felice Bombaci, Rita O. Christensen, Bahija Gouimi, Mina Daban, Nigel B. Deekes, Jennie Bradley

P138 | KTH - Royal Institute of Technology
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Andreas Hober, Fredrik Edfors, Erik Niks, Pietro Spitali, Cristina Al-Khalili Szigyarto

P140 | OPEN Health Medical Communications
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Ana Hernandez
Nirali Kotowsky, Ran Gao, David Singer, Elizabeth M Garry, Amanda K Golembesky

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Gavin McDonough
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Terese Johansson
Pauline Leslie, Arwa Shurrab, Morihiro Sakata, Rosie Dawes

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Alessandra CAMERINI
Alessandra Zatti, Ermanno Rizzi, Elena Bruno, Manuela Battaglia

P144 | Dutch Pituitary Foundation / Endo-ERN
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Johan de Graaf
Prof. dr. Martine Cools, Prof. dr. Marta Korbonits, Prof. dr. Alberto Pereira, Prof. dr. Olaf Hoirt, Johan de Graaf

P145 | Syneos Health
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Maryna Kolochavina
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P146 | Association Française du Diabète Insipide (AFDI)
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Buono Suzie
Laurent Hélène and Poly Marie-sophie

P147 | Vifor Fresenius Medical Care Renal Pharma
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Dijana Krafcsik
Peter Rutherford (MD), Dieter Götte (MD)

P148 | University of Calgary/ Canadian Neuromuscular Disease Registry
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Linh Vu
Hodgkinson V, Vu L, M'Dahoma S, Lounsberry J, Brais B, Campbell C, Johnston W, McCormick A, Nguyen CT, O'Ferrall E, Oskoui M, Korngut L and the CNDR Investigator Network.

P149 | Fundación Española para el Estudio y Terapéutica de la Enfermedad de Gaucher y otras lisosomales (FEETEG)
TRAZELGA Project: Preliminary results of the prospective national-base multicenter study to standardize the follow-up of type 1 Gaucher disease patients treated with Eliglustat.
Irene Serrano Gonzalo
López de Frutos L, Cebolla JJ, Mora E, Lahoz C, Fernández-Galán MA, Morales-Conejo M, Delgado-Mateos F, Calle-Gordo V, Vitoria-Miñana I, Correcher-Medina P, Rodríguez-Fernández A, Hernández-Rivas JM, García-Frade J, Ibarretxe-Gerediaga D, Villalón-Blanco L, Tormo-Díaz MM, Ruíz-Guinaldo MA, Mora-Castera E, Noya-Pereira MS, López-Dupla M, Balanzat-Muñoz J, Fernández-Cofrades E, Lozano-Almela ML, Pérez de León, Molero-Labarta T, Fernández-Canal MC, Pérez-Montaña A, Andrade-Campos M, Giraldo P.

P151 | European Paediatric Translational Research Infrastructure
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Donato Bonifazi
Mariangela Lupo, Valeria Pignataro, Giovanni Migliaccio, Adriana Ceci

P152 | Cystinosis Ireland
Worldwide Cystinosis Community Advisory Board
Denise Dunne
Denise Dunne, Anne Marie O'Dowd, Rob Camp

P153 | CERGAS SDA Bocconi
The estimation of health state utility values in rare diseases: overview of the existing techniques.
Michela Meregaglia
Elena Nicod, Michael Drummond

P154 | Centre for Research on Health and Social Care Management, SDA Bocconi University
Using PROMs in HTA for Rare Disease Treatments
Amanda Whittal
Elena Nicod, Michela Meregaglia, Mike Drummond

P155 | Fondazione Policlinico Universitario «A. Gemelli» IRCCS
Antithrombotic therapy in subjects with hereditary hemorrhagic telangiectasia: update of prospective data from a Multidisciplinary group
Fabiana Agostini
Eleonora Gaetani, Angelo Porfidia, Igor Giarretta, Annalisa Tortora, Daniela Feliciani, Luigi Di Martino, Antonio Gasbarrini, Roberto Pola

P157 | Dutch CF Foundation
The Cystic Fibrosis Community Advisory Board (CF-CAB) – how we measure our successes
Marja Nell
Rob Camp, Hilde de Keyser

P158 | EURORDIS
The Value of Patient Engagement in Research Design: The EURORDIS Patient-Led Community Community Advisory Boards (CABs)
Rob Camp
François Houÿez

P159 | Genetic Alliance UK
Priorities for research in primary mitochondrial disease: patients' and clinicians' Top 10.
Amy Hunter
The Steering Group of the Mitochondrial Disease Priority Setting Partnership

P160 | LEO Pharma
Co-Creation from the Get-Go - Understand the Known and Unknown needs
Lasse Funch Jacobsen
Lasse Funch Jacobsen

P161 | ERN-EYE
How to welcome visually impaired people to the hospital? ERN-EYE Educational video for healthcare professionals
Claire Edel

P162 | Newcastle University
Expert advice to improve clinical trials: from TACT to ACT
Joanne Lee
Volker Straub, Annamaria De Luca, Cathy Turner, Victoria Hedley

P163 | AKU Society
The first effective treatment for AKU: A collaborative, patient centric effort
Ciarán Scott
Prof Lakshminarayan Ranganath, Nick Sireau

P164 | Andalusian Network for Advanced Therapies - Andalusian Public Foundation Progress and Health
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Ivana Kildsgaard
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P167 | Ultragenyx Pharmaceutical Inc.
The MPS VII Disease Monitoring Program (DMP) is a novel, longitudinal, cohort program with rigor beyond a traditional registry
Deborah Marsden, MD
Robert Hostutler MS, Tricia Cimms MPH

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P168 | Sorbonne Université - Inserm UMRS 974, Center of Research in Myology, Institut de Myologie, G.H. Pitié-Salpêtrière Paris, France
Treatabolome: a rare diseases treatment awareness project
Antonio Atalaia
Antonio Atalaia, Rachel Thompson, Alberto Corvo, Leigh Carmody, David Pisicia, Leslie Matalonga, Alfons Macaya, Angela Lochmuller, Bertrand Fontaine, Birte Zurek, Carles Hernandez-Ferrer, Carola Reinhard, David Gómez-Andrés, Gulcin Gumus, Jean-François Desaphy, Katherine Schon, Katja Lohmann, Matthew Jennings, Matthis Synofzik, Olaf Riess, Rabah Ben Yaou, Teresinha Evangelista, Thiloka Ratnaike, Virginie Bros-Facer, Rita Horvath, Patrick Chinnery, Steven Laurie, Holm Graessner, Peter Robinson, Hanns Lochmüller, Sergi Beltran, Gisèle Bonne

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P170 | Medical Informatics Group (MIG), Frankfurt, University Hospital, Theodor-Stern-Kai 7, 60590 Frankfurt, Germany
The internet platform EXABO – from beta to full version
Désirée Walther
Désirée Walther, Olivia Steinmann, Jannik Schaaf, Thomas Wagner and Holger Storf

P171 | Biological Research Center Margarita Salas
Targeting β2-adrenergic receptors as a new strategy for von Hippel-Lindau disease
Angel M Cuesta
Albiñana V, I. de Rojas-P, Gallardo-Vara E, Recio-Poveda L, Villar Gómez de Las Heras K, Aguirre DT, and Botella LM.

P172 | KU Leuven
How to define the value of orphan drugs? A comparative analysis of value assessment frameworks across Europe (EUROVAF)
Alessandra Blonda
Isabelle Huys, Yvonne Denier, Steven Simoens

P173 | University of Leicester
Project HERCULES: The Challenges of Estimating Multi-State Model Transitions in Rare Diseases: Creating a Natural History Model for Duchenne Muscular Dystrophy (DMD)
Jonathan Broomfield
Broomfield J, Hill M, Crowther MJ, Larkindale J, Guglieri M, Godfrey J, Chandler F, Abrams KR

P177 | The Finnish Association of People with Physical Disabilities
The comprehensive musculoskeletal post-acute ICF core set as a tool to formulate a questionnaire and internally consistent sum variables describing people with skeletal dysplasia
Hanna Hyvönen
Hanna Hyvönen, Heidi Anttila, Susanna Tallqvist, Minna Muñoz, Sanna Leppäjoki, Antti Teittinen, Outi Mäkitie, Sinikka Hiekkala

P179 | Dolon
Managing clinical and economic uncertainty in the value assessment of innovative gene and cell therapies – adjustments to health technology assessments and innovative payment models
Darren Callanan
Richard Sear, Karolina Hanslik, Marc Dziasko

P180 | Comenius University in Bratislava, Faculty of Pharmacy, Dpt. of Pharmacology and Toxicology and Slovak aliance for rare diseases
How accessible are orphan medicinal products in Slovakia?
Tatiana Foltanova
Gabriela Hrckova

P181 | Consejo Superior de Investigaciones Científicas (CSIC) (National Research Council Spain)
Propranolol, an accessible, affordable, and available orphan drug for the von Hippel-Lindau disease.
Luisa Maria Botella Cubells
Albiñana, V, de las Heras K, Gonzalez B, Jimenez R, Aguirre, D, Cuesta AM

P182 | KU Leuven
Methodological Quality of Budget Impact Analyses for Orphan Drugs: An Updated Review
Abdallah Khadidja
Huys Isabelle, Claes Kathleen, Simoens Steven

P183 | Partners4Access
Driving patient access for treatments targeting rare genetic disorders in the Middle East
Ciaran Cassidy
Akshay Kumar

P184 | European Confederation of Pharmaceutical Entrepreneurs (EUCOPE)
A 20th anniversary analysis of the EU OMP Regulation: achievements and future outlooks
Vittoria Carraro

P185 | Centre for Research on Health and Social Care Management, SDA Bocconi University
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Amanda Whittal
Elena Nicod

P186 | Metabolic Support UK
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Lindsay Weaver
Lindsay Weaver, Pushpa Hossain, Joanne Byrne & Josie Godfrey

P187 | Chinese Organization for Rare Disorders
China Rare Disease Drug Accessibility Report 2019
Kevin Huang
ERIC SHAO, EILEEN LI, GRACE WANG, MIRROR ZHU, LEI XIAO, MAKBULE TOP, QI SUN

P188 | Sarepta Therapeutics
Health-related quality of life (HRQoL) and economic burden of Duchenne Muscular Dystrophy (DMD): a systematic literature review
Annika Bergman
J. Iff, C. McCloskey, C. Fidler, N. Goemans

P189 | Global Market Access Solutions
Estimating the broader fiscal impact of rare diseases using a public economic framework: A case study applied to acute hepatic porphyria (AHP)
Mark Connolly
Saswat Panda, Julien Patris

P190 | Unit of Pharmacoepidemiology & Pharmacoeconomics, Department of Pharmacy, University of Groningen
Estimating the broader fiscal impact of rare diseases using a public economic framework: A case study applied to hereditary transthyretin-mediated (hATTR) amyloidosis
Mark Connolly
Julien Patris, Saswat Panda, Bouke P.C. Hazenberg

P193 | DEBRA Ireland
Economic Burden of Epidermolysis Bullosa: Evidence from Ireland
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Darragh Flannery, Claire Doyle, Avril Kennan, Fiona Aherne

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The European Cystic Fibrosis Society Patient Registry's Data Quality programme.
Andreas Jung
Van Rens J., Fox A., Krasnyk M., Orenti A., Zolin A., Jung A., Naehrlich L.

P195 | Casimir
Evidence of content validity of the Duchenne video assessment scorecards from a Delphi panel study
Marielle Contesse
Linda Lowes, Michelle K. White, Laura Dalle Pazze, Christine McSherry, Mindy Leffler

P196 | Aparito
PROvide: Video based Patient Reported Outcomes for Sanfilippo Disease: a new and innovative approach to record and measure disease post gene therapy.
Dr Elin Haf DAVIES
C O'Neil, S Parker, M Leffer

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Enrico Manzini
Jon Garrido-Aguirre, Alexandre Perera-Lluna

P199 | Aparito
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Cécile Ollivier
Elin Haf Davies, Pip Griffiths

P200 | Frambu resource centre for rare disorders
Development of SIBS-online: an internet-based group intervention for siblings and parents of children with rare disorders
Torun Marie Vatne
Lauren Kelada, Silje Systad and Krister W. Fjermestad

P201 | Stichting Shwachman Syndrome Support Holland
Global semantic interoperability a tool to ensure full participation in society.
Rob Braamburg
Petra Poulissen, Elmas Citak, Tessa Risch, Liesbeth Siderius

P202 | Novartis Pharma AG
Virtual Reality: An Innovative approach to sharing learnings with Health Care Professionals on Cytokine Release Syndrome and neurological events after Chimeric Antigen Receptor T Cell Therapy (CAR-T) Infusion
Simona Paratore

P203 | IRCCS Istituto Ortopedico Rizzoli
Establishing the first pan-European Registry for Rare Bone and Mineral Disorders
Marina Mordenti
Marina Mordenti, Ines Alves, Rebecca Skarberg, Ondrej Soucek, Marco Ross, Corinna Grasemann, Kassim Javaid, S. Faisal Ahmed, Agnès Linglart, Klaus Mohnike, Wolfgang Högler, Luca Sangiorgi, and Natasha Appelman-Dijkstra

P204 | University Hospital of Frankfurt, Department of Paediatric Surgery and Paediatric Urology
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Andrea Schmedding
Udo Rolle, Jessica Vasseur, Holger Storf and the KiRaFe-Group

P205 | Meditech Media
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Florian Lipsmeier
Cedric Simillion, Atieh Bamdadian, Anne Smith, Scott A. Schobel, Rosanna Tortelli, Filipe B. Rodrigues, Lauren M. Byrne, Edward J. Wild, Michael Lindemann

P206 | Ada Health GmbH
Just how rare are rare diseases on Ada, a medical symptom checker?
Vanessa Lemarié
Paul Wicks, Simon Ronicke, Fiona Pick

P207 | Medical Informatics Group, University Hospital Frankfurt
SAVE – An online tool to improve diagnosis and therapy of patients with cystinosis as an exemplary approach for rare diseases
Jessica Vasseur
Froschauer S, Göbel J, Boulesteix A-L, Erler J, Herzig N, Holla H, Knerr C, Koeppl C, Niessl C, Passow M, Priglinger C, Steidle G, Treikauskas U, Wolff S, Weitzel D, Vill K, Hohenfellner K, Storf H

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Giacomo Frisoni
Gianluca Moro, Antonella Carbonaro

P210 | Sant Joan de Déu Research Foundation
Share4Rare: a collaborative platform for rare diseases
Sara Hernández-Ortega
Dimitrios Athanasiou, Nicholas Brooke, Inma Chapí, Anaïs Lecorvec, Avril Palmeri, Alex Perera, Bettina Ryll, Begonya Nafria

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Angela Wilson
Toni Mathieson, Shaun Bolton, Jackie Imrie

P212 | European Reference Nertwork Rare Eye Diseases (ERN-EYE)
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Agnė Kručaitė
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P214 | Centre for Rare Diseases Tübingen (CRDT), University Hospital Tübingen (UKT), Calwer Str. 7, 72076 Tübingen, Germany
ORPHAcoding RD Patients in a Rare Disease Centre - The Tübingen Solution
Holm Graessner
Jutta Eymann, Vera Belka, Maria Gulde

P215 | Vrije Universiteit Brussel
Beliefs of people with a rare disease about how active a role they can play when consulting a specialist
Katlijn Sanctorum
An Jacobs

P216 | Geneva University Hospitals
Preliminary Evaluation of a mHealth Coaching Conversational Artificial Intelligence (AI) for the Self-Care Management of People with Sickle-Cell Disease (SCD)
David-Zacharie Issom
Jessica Rochat, Gunnar Hartvigsen, Christian Lovis

P217 | Rare diseases Sweden
Social media training by patient organisations for patient organisations
Britta Berglund
Jana Bergholtz and Mildred Lundgren

P218 | Hackensack University Medical Center
A Data Analytics Approach to Rare Disease Discovery
Kenneth Lieberman, MD

THEME 7: RARE DISEASE PATIENT GROUPS INNOVATIONS

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Yukiko NISHIMURA
Shun Emoto, Masatoshi Iwasaki, RDD Japan team, and RDD Japan Secretariat office

P220 | Planet 5p
Planet 5P Project. A Cri Du Chat Syndrome Global Community
Cristina Bel Fenellós
Carolina Nicolás Lozano, Margarita Pajón Naranjo

P221 | Genetic Alliance UK
Developing Rare Resources for Scotland: Utilising methods of co-production to meet the information needs of people with rare, genetic or undiagnosed conditions and the health professionals involved in their care.
Natalie Frankish

P222 | Cystic Fibrosis Europe
Improving communication about clinical trials in Cystic Fibrosis: a starting point for plain language summaries
Hilde De Keyser (1)
Emmanuelle Bardin, Jill Bonjean, Fiona Dunlevy, Rebecca Brendell, Lorna Allen.

P223 | Deutschsprachige Selbsthilfegruppe für Alkaptonurie e.V.
Setting up an infra-structure of healthcare for Alkaptonuria in Germany by dissemination and networking
Leona Wagner
Harald Wilke, Marion Hoyer, Gerhard Meng, Konstantinos Kolokotronis, Stephan vom Dahl, Anibh Das, Athanasia Ziagaki

P224 | Salla Treatment and Research Foundation
The use of social media to create a foundation and drive research on rare diseases
Ann Persson

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Christine Mutena

P227 | CML Advocates Network
CML Patients Advocates Network implementation of the Community Advisory Board (CAB) Model
Celia Marín

P228 | MPNErare- MPNE's subnetwork for rare Melanomas
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Bettina Ryll
Bettina Ryll, Gilly Spurrier, Violeta Astratinei, Fredrik Östman, Iain Galloway, Andrew Evans, Erik Näsman, Oskar Näsman, Dick Plomp, Audrey Woraker, Martina Rooijakkers, Ioana Dogaru and Jo Gumbs

P231 | JG Zebra Consulting
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Emily Crossley
Fleur Chandler, Josie Godfrey, Keith Abrams, Jill Carlton, Juliet Mumby-Croft, Jamie O'Hara, Philip Powell,

P232 | Medics4RareDiseases
Consistent follower engagement on social media can grow your audience and raise awareness of rare diseases
Dr Lucy McKay
Jo McPherson

P233 | Digimevo
Improving patient communication and education using POC (Point of Care) Channel
Xavier Lleixà
Lleixa X, Panisello J, Carro A, Fonfría M, Solé M

P234 | CIBERER
CIBERER experience: Giving Patients a Seat at the Table
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Beatriz Morte

P235 | Findacure
Building a set of training resources to help rare disease patient groups form, grow, and maximize their impact.
Mary Rose Roberts
Mary Rose Roberts, Philippa Norman, Laura Thompson-Harper

P236 | Swedish Association of Chronic Oedema (Svenska Ödemförbundet)
Lympho-logical thinking in self management of primary lymphoedema and other rare diseases involving lymphoedema
Margareta Haag
Helena Janlöv Remnerud

P237 | San Raffaele Scientific Institute
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DIA is where science, healthcare and regulation unite. Gathering the brightest minds around the globe, we build a network that is multidisciplinary and impactful. Together, we foster innovation and improve the wellbeing of patients worldwide. Engage with DIA through our unparalleled membership network and professional development opportunities.

EJP

The European Joint Programme on Rare Diseases (EJP RD) brings over 130 institutions (including all 24 ERNs) from 35 countries to create a comprehensive, sustainable ecosystem allowing a virtuous circle between research, care and medical innovation.

EMA

The European Medicines Agency (EMA) is a decentralised agency of the European Union (EU). The Agency is responsible for the scientific evaluation, supervision and safety monitoring of medicines in the EU. EMA protects public and animal health in EU Member States, as well as the countries of the European Economic Area, by ensuring that all medicines available on the EU market are safe, effective and of high quality.

EURORDIS

Come and visit the EURORDIS stand to find out the latest about our projects and activities, see our publications and speak to our team. The organisers behind ECRD, EURORDIS-Rare Diseases Europe is a unique, non-profit alliance of 917 rare disease patient organisations from 72 countries.

EUROPEAN MEDICAL JOURNAL

The EMJ, is an independent, open-access eJournal dedicated to delivering first-class insights into ground-breaking changes, and advancements in medicine. Spanning eighteen therapeutic areas, including Innovations, Cardiology, Oncology, and more, each journal provides the reader with the latest medical congress highlights, abstract reviews, and peer-reviewed articles to name but a few of its wide content selection.

EUROPEAN REFERENCE NETWORKS

The European Reference Networks (ERNs) gather doctors and researchers with high expertise in the fields of rare or low-prevalence and complex diseases. They are “virtual networks” which discuss the diagnosis and the best possible treatment for patients from all over Europe. 24 ERNs were launched in 2017, involving more than 900 highly specialised healthcare teams, located in more than 300 hospitals in 26 European countries.

EXHIBITORS

GLOBAL COMMISSION

The Global Commission is a multidisciplinary group of experts from around the world, co-chaired by Takeda, Microsoft and EURORDIS-Rare Diseases Europe, who have brought their creativity, technological expertise and passion to accelerate the time to diagnosis for children with a rare disease. Our vision is a clear path to a timely, accurate diagnosis for children around the world.

IRDIRC

The International Rare Diseases Research Consortium (IRDiRC) unites national and international governmental and non-profit funding bodies, companies (including pharmaceutical and biotech enterprises), umbrella patient advocacy organizations, and scientific researchers to promote international collaboration and advance rare diseases research worldwide.

OPENAPP

OpenApp develop patient registries for patient reported and clinical data. Our platform has been configured for many therapeutic areas, generating Real World Evidence to support advocacy, research, and pharmacovigilance. Stop by our virtual booth to learn more and speak to a representative.

ORCHARD THERAPEUTICS

Orchard Therapeutics is a global gene therapy leader dedicated to transforming the lives of people affected by genetic diseases through the development of innovative, potentially curative gene therapies. Our ex vivo autologous gene therapy approach harnesses the power of genetically-modified blood stem cells and seeks to correct the underlying cause of disease in a single administration.

ORPHANET

Orphanet (www.orpha.net) is a unique resource, gathering and improving knowledge on rare diseases. Orphanet aims to provide high-quality information on rare diseases, and ensure equal access to knowledge for all stakeholders. Orphanet also maintains the Orphanet rare disease nomenclature (ORPHAcode), essential in improving the visibility of rare diseases in health and research information systems.

RARE 2030

Rare 2030 is a foresight study that gathers the input of a large group of patients, practitioners and key opinion leaders to propose policy recommendations that will lead us to improved policy and a better future for people living with a rare disease in Europe. This a two year project that will end in a presentation to parliament at the end of 2020 with recommendations on the most critical areas needing sound policy.

RARE BAROMETER

Rare Barometer is the EURORDIS evidence-based advocacy programme that aims to transform rare disease patients' opinions and experiences about topics that directly affect them into figures and facts. It connects researchers and policymakers to questions that matter to rare disease patients. Rare disease patients can register for our programme and take part in our surveys and studies.

RARE REVOLUTION MAGAZINE

Rare Revolution Magazine provides exceptional articles of interest to the rare disease community. You will find compelling voices from rare disease advocates and patients, articles from clinical, research and pharmaceutical teams and the latest in 'RARE' advancements.
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SAREPTA

Armed with the most advanced science in genetic medecine, we are in a daily race to rescue lives otherwise stolen by rare disease. At Sarepta, everyday is another twenty-four hours to stand up for patients, advance technology, challenge convention, and drag tomorrow into today.

SBONN

SBONN was founded in 2014 by umbrella organisations and networks representing people living with rare diseases in Denmark, Finland, Iceland, Norway and Sweden. SBONN aims to promote the exchange of ideas, knowledge, learning and understanding of living with a rare disease and/or disability across national borders in the Nordic region.

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