HYBRID ecrd

ACTION WITHIN REACH

PIONEERING SOLUTIONS FOR RARE DISEASES

HYBRID 15 & 16 MAY 2024

12th European Conference on Rare Diseases and Orphan Products

DIGITAL PROGRAMME

Under the auspices of

belgium24.eu

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

#ECRD2024

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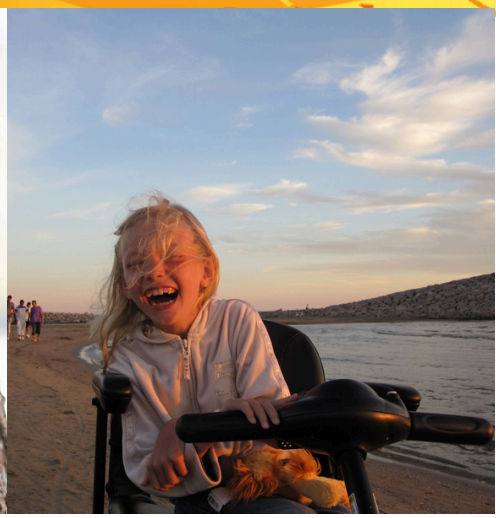

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All times listed are Central European Summer Time (CEST)

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.

"If we were capable of doing what we did for cancers, we can do it for rare diseases."

STELLA KYRIAKIDES, European Commissioner for Health and Food Safety

WHY DOES EUROPE NEED TO TAKE ACTION ON RARE DISEASES, NOW?

ECRD is a great opportunity to translate the current political momentum into comprehensive actions for the EU's next cohort of policymakers and leaders. The ECRD 2024 programme will do this by addressing the key policy areas that form part of a broader policy framework. The conference will culminate in the output of a co-created, open letter to the EU Institutions and country leaders where expectations of the community are clearly spelled out and conveyed to decision-makers in an unequivocal manner, leaving a solid legacy for the future EU leadership. You have the opportunity to be part of this significant moment!

MESSAGE FROM THE CO-CHAIRS

Adéla Odrihocká

Patient Advocate, Rare Diseases Czech Republic

Claas Röhl

Chair, ProRare Austria

Eva Schoeters

Director, RaDiOrg - Rare Diseases Belgium

DEAR PARTICIPANTS,

We are delighted to welcome you on behalf of conference organisers **EURORDIS-Rare Diseases Europe**, co-organisers **Orphanet**, and all other conference partners to the 12th European Conference on Rare Diseases & Orphan Products 2024, taking place in hybrid format, both at The Square in Brussels and online, on 15-16 May 2024. Globally recognised as the largest, patient-led rare disease policy event, **ECRD 2024** has been organised under the auspices of the **Belgian Presidency of the Council of the European Union**. The overarching theme for this year's conference is **Action Within Reach: Pioneering Solutions for Rare Diseases.**

With the European elections approaching, at the inception of a new term for the **European Parliament** and the **European Commission**, the **12th ECRD** becomes a key driver in shaping policies to address the unique needs of people with rare diseases and their families over the next five years. The **ECRD**'s value is underscored by its alignment with EURORDIS' priorities for the EU elections, evident in its comprehensive six-session agenda.

What sets the **ECRD** apart is its integration into broader rare disease policy and healthcare ecosystem efforts. Rooted in the recommendations of the Rare 2030 Foresight Study, the **ECRD** continues to build on this comprehensive review of the strategy for rare diseases and shape the thinking of policymakers and the community, encouraging a more streamlined and proactive approach to addressing the unmet needs of people living with rare diseases and the persisting inequalities across Europe.

This conference will focus on identifying the most critical priorities and areas for improvement. By bringing together stakeholders from across Europe, we aim to build a multi-stakeholder consensus on a comprehensive approach to rare disease policy that can provide a footprint for the next EU legislative cycle.

In addition to exploring the most impactful policy directions, this conference will be the occasion to showcase the dynamic ideas coming from different stakeholders across Europe. With a bottom-up approach to designing the path forward, our participants will hear about different initiatives at the national and local level of the advancements made from national and regional strategies, as well as public-private initiatives. **ECRD 2024** will explore how to build on these initiatives and create a comprehensive European approach to rare diseases.

Finally, we will demonstrate how rare diseases are transforming health care and driving new approaches to public health. This will include rare cancers and precision medicine, early diagnosis and newborn screening and digital/data.

This year, the conference will feature closed captioning in **12 languages** to accommodate our diverse international attendees. Available languages include **Spanish, Portuguese, German, French, Italian, Polish, Romanian, Dutch, Greek, Hungarian, Czech and Danish**. This service is part of our commitment to making the conference accessible and inclusive for all participants.

We are delighted that you are joining **ECRD 2024** and hope you enjoy the conference as an unrivalled opportunity to network and exchange invaluable knowledge with all stakeholders in the rare disease community from over 45 countries around the world.

Best regards,

The ECRD 2024 Programme Co-Chairs

PROGRAMME AT A GLANCE

All times are Central European Summer Time (CEST)

The ECRD is the largest, patient-led, rare disease policy-shaping event held in Europe. By bringing together people with rare diseases and patient advocates, policy makers, healthcare industry representatives, clinicians, regulators and Member State representatives, EURORDIS harnesses the power of this extensive network to shape goal-driven rare disease policies of the future.

With over 600 participants, the Conference is an unrivalled opportunity to network and exchange invaluable insights within the rare disease community. Through collaborative efforts, these discussions culminate in clear policy recommendations that can influence both EU and national policies.

Get ready for ECRD 2024, a fully hybrid conference taking place on 15 & 16 May 2024 online and at The Square in Brussels!

ECRD DAY 1 WEDNESDAY 15th MAY 2024

08:45 - 09:30	**MORNING CHECK-IN**
09:30 - 11:00	**OPENING PLENARY SESSION**
11:00 - 11:30	**COMFORT BREAK**
11:30 - 12:30	**POSTER PITCH**
12:30 - 14:00	**LUNCHTIME**
14:00 - 15:30	**PARALLEL SESSIONS** Revolutionising Funding Strategies for Breakthrough Therapies in Rarer Diseases No Health without Mental Health! Let's Co-create a Mentally Healthy Toolkit
15:30 - 16:10	**COMFORT BREAK**
16:10 - 17:15	**SMALL GROUP DISCUSSIONS**
17:15 - 18:00	**HIGHLIGHTS HUB**
18:00 - 19:30	**NETWORKING RECEPTION**

PROGRAMME AT A GLANCE

All times are Central European Summer Time (CEST)

ECRD DAY 2 THURSDAY 16th MAY 2024

08:30 - 09:00	**MORNING CHECK-IN**
09:00 - 09:45	**OPENING PLENARY SESSION**
09:45 - 10:45	**POSTER AWARDS AND POSTER PITCH**
11:00 - 11:30	**COMFORT BREAK**
11:30 - 13:00	**PARALLEL SESSIONS**  The Path Forward for Equitable Diagnosis  Achieving Full Reach: Overcoming the Last Challenges to Access Highly Specialised Care
13:00 - 14:00	**LUNCHTIME**
14:00 - 15:30	**PARALLEL SESSIONS**  Innovative Therapies, Unequal Access: Bridging the Gap for Rare Disease Treatments  National Plans: Exchanging Best Practices to Forge a Unified European Response to Rare Disease
15:30 - 16:15	**COMFORT BREAK**
16:15 - 16:45	**HIGHLIGHTS HUB**
16:45 - 17:30	**CLOSING PLENARY SESSION**


RESEARCH & INNOVATION


MENTAL HEALTH & WELLBEING


DIAGNOSIS & SCREENING


ACCESS TO HIGHLY SPECIALISED CARE


ACCESS, AVAILABILITY AND AFFORDABILITY OF TREATMENTS


RARE DISEASE NATIONAL PLANS

A HYBRID CONFERENCE

JOINING THE CONFERENCE REMOTELY

- Using the event's virtual platform, you can join ECRD from anywhere you wish, with the possibility to re-watch on-demand sessions at your convenience.
- Direct from your desk - participate in sessions run by leading experts built around 6 topics. Over 50 chairs and speakers will lead interactive sessions and be available to answer questions.
- Professional moderators dedicated to online participants will ensure your voice is heard throughout the conference and provide you with equal opportunity to engage.
- Special online registration fees are available for all stakeholder groups to take part!
- Access to hundreds of multimedia ECRD posters.

The Faculty of Pharmaceutical Medicine of the Royal Colleges of Physicians of the United Kingdom has approved this conference. The conference will be honoured with 11.75 CPD credits.

CLOSED CAPTIONING & TRANSLATIONS

Closed captioning will be available for all ECRD sessions in 13 languages.

Languages include English, French, Czech, Danish, Dutch, German, Greek, Hungarian, Italian, Polish, Portuguese, Romanian and Spanish.

NETWORKING EVENTS

The online conference platform has been designed with user-friendliness in mind to provide an accessible and engaging virtual experience for all participants.

YOU WILL BE ABLE TO:

- Join the General Chat popping up on all the pages - you can either send a message to everyone or a direct one to a specific participant.
- Look up attendees by their Name/ Company/Registration Type/Country and request a call in the **Meet the Participants** section. On the same page, you will also find a carousel of recommended contacts based on the number of common interest fields you have listed on your registration. If you haven't yet provided these, we invite you to do so under **My Profile.**
- Add your key commitments from the conference to our **Virtual Commitment Wall** and support the rare disease community on a personal, community or organisational level. Share your statement to contribute to the legacy ECRD 2024 is leaving and represent your commitment to translating **intention into action**!

Use the hashtags and post on your favorite social media, you'll appear on the **Social Media Wall** and in the homepage (#ECRD2024)!

Alternatively, take a selfie, GIF or boomerang in our **Virtual Photo Booth** to show everyone you attended ECRD 2024!

IN PARTNERSHIP WITH

Under the auspices of

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WITH CO-FUNDING FROM

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European Brain Council

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GUÉRIR LES MALADIES GÉNÉTIQUES

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INTERNATIONAL RARE DISEASES RESEARCH CONSORTIUM

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NHS Foundation Trust
THE John Walton MUSCULAR DYSTROPHY RESEARCH CENTRE
Newcastle University

MEDICS 4 RARE DISEASES

Newcastle University
Centre for Rare Disease

raremínds
Mental Health for the Rare Disease Community

SCREEN 4CARE

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for rare or low prevalence complex diseases
Network
Craniofacial anomalies and ear, nose and throat disorders (ERN CRANIO)

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Epilepsies (ERN EpiCARE)
Coordinator
Hospital Sant Joan de Déu — Spain

European Reference Network
for rare or low prevalence complex diseases
Network
Hematological Diseases (ERN EuroBloodNet)

European Reference Network
for rare or low prevalence complex diseases
Network
Urogenital Diseases (ERN eUROGEN)

European Reference Network
for rare or low prevalence complex diseases
Network
Neuromuscular Diseases (ERN EURO-NMD)
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Assistance Publique - Hôpitaux de Paris, Hôpital Pitié-Salpétrière — France

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for rare or low prevalence complex diseases
Network
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European Reference Network
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Genetic Tumour Risk Syndromes (ERN GENTURIS)

ERN GENTURIS
With every diagnosis we can help an entire family

European Reference Network
for rare or low prevalence complex diseases
Network
Heart Diseases (ERN GUARD-HEART)
Coordinator
Academic Medical Center Amsterdam — The Netherlands

ITHACA

European Reference Network

ERN RARE-LIVER

European Reference Network

ERN ReCONNET

rita

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SKIN

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

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

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for rare or low prevalence complex diseases
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EUROPEAN REFERENCE NETWORK ON RARE BONE DISEASES

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COMMITTEES

PROGRAMME COMMITTEE **MEMBERS**

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Inserm-Orphanet

Anne- Sophie Lapointe
French Ministry of Solidarity and Health

Ariane Weinman
EURORDIS

Claas Rohl
NF Kinder / EUPATI Austria / Alliance of Allianz der österreichischen Patient

Dorica Dan
EURORDIS

Stefan Zivkovic
National Organisation for Rare Diseases of Serbia (NORBS) / Bringing Europeans Together Association Serbia

Yann Le Cam
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Matt Bolz-Johnson
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EURORDIS

Simona Bellagambi
UNIAMO

Simone Boselli
EURORDIS

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EURORDIS

COMMITTEES

OUTREACH COMMITTEE **MEMBERS**

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Allianz Chronischer Seltener Erkrankungen

Ulrike Holzer
Pro Rare Austria

Birthe Byskov Holm
Rare Diseases Denmark

Peter Saltonstall
Nord National Organization for Rare Disorders

Cor Oosterwijk
VSOP - Vereniging Samenwerkende Ouder En Patiëntenorganisaties

Ramaiah Muthyala
Indian Organization for Rare Diseases

Simona Bellagambi
UNIAMO

Anna Arellanesová
Rare Diseases Czech Republic

Yukiko Kuroda Nishimura
Shun Emoto
Japan Patient Association

Paula Gonçalves
Rare Diseases Portugal

Vicky McGrath
Rare Diseases Ireland

Charalampos Papadopoulos
Cyprus Alliance for Rare Disorders

EVENTS AROUND THE CONFERENCE
APRIL - MAY 2024

PODCAST EPISODES

Take a deep-dive into some of the key conference themes

See the conference website to listen!

THOUGHT LEADER SESSIONS - WATCH RECORDINGS HERE!

Harnessing Digital Solutions for Rare Diseases
Thursday 25th April 2024
12.00 - 13.00 CET
Click here for the Session Programme

Collaborating for Change - Transforming Rare Disease Outcomes through Public-Private Partnerships
Friday 3rd May 2024
15.00 - 16.00 CET
Click here for the Session Programme

The Global Commission to End the Diagnostic Odyssey for Children with a Rare Disease
Tuesday 7th May 2024
14.00 - 15.00 CET
Click here for the Session Programme

SATELLITE MEETINGS

Satellite Meeting - Organised by the Romanian National Alliance for Rare Diseases – RONARD
Monday, 13th May 2024
15.00-17.00 CET
See the conference website for updates

ECRD DAY 1 WEDNESDAY 15th MAY 2024

PLENARY SESSIONS

Facilitated by:

Tamsin Rose
Professional Facilitator

Gerrit Heijkoop
Professional Facilitator

OPENING PLENARY SESSION

9.30 - 11.00

WELCOME REMARKS BY FACILITATORS

KEYNOTE ADDRESSES

Avril Daly
President, EURORDIS- Rare Diseases Europe

Virginie Bros-Facer
Chief Executive Officer, EURORDIS-Rare Diseases Europe

Frank Vandenbroucke
Minister of Health and Social Affairs, Belgium

PAST PARTICIPANT TESTIMONY

Lucy McKay
CEO, Medics4RareDiseases

INTERACTIVE MUSICAL INTERLUDE

NEWCOMER TESTIMONY

Stefan Joris
Chair, Association Muco Vereniging

PROGRAMME OVERVIEW

Ana Rath
Director, Orphanet

CLOSING REMARKS BY FACILITATORS

ADDITIONAL PLENARY SESSIONS

Running throughout the day

11.30 - 12.30

LEARN AND PITCH YOUR IDEAS IN THE POSTER PITCH

Learning from each other is an integral part of the ECRD experience. Top-scoring poster authors will provide short overviews of their research, studies or publications to whet your appetite and entice you to delve deeper into the posters showcased throughout the conference.

This interactive pitch session will be followed by a poster scavenger hunt, facilitated both online and in-person, with prizes to be won via the interactive online conference platform!

16.10 - 17.15

JOIN SMALL GROUP DISCUSSIONS

This is your chance to have your say on which topics should be discussed during this session! Share your ideas by email at: events@eurordis.org.

During the Day 1 Plenary sessions, you will have the opportunity to vote for your favourite topics to be discussed. Share your ideas using **this form.**

During this session, simply select from the confirmed topics and join the relevant group for an informal exchange. No professional moderation provided. Sessions will be duplicated both online and in-person giving everyone the opportunity to connect with others around topics that are important to you.

17.15 - 18.00

HIGHLIGHTS HUB

With the help of Geoff Case, Digital Editor and Senior Associate at RARE Revolution Magazine, we'll discover the most important insights and highlights from Day 1 of ECRD. This session will feature short interviews with key speakers and session chairs, summarising the most important discussions and policy outcomes. This interactive catch-up will also provide opportunities for you to share your own perspectives.

ECRD DAY 2 THURSDAY 16th MAY 2024

PLENARY SESSIONS

Facilitated by:

Tamsin Rose
Professional Facilitator

Gerrit Heijkoop
Professional Facilitator

OPENING PLENARY SESSION

9.00 - 9.45

OPENING REMARKS BY FACILITATORS

KEYNOTE ADDRESSES

Eva Schoeters
Director, RaDiOrg

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

Sofie Skoubo
Paralympic Athlete and PhD Student

ADDITIONAL PLENARY SESSIONS

Running throughout the day

9.45 - 10.45
POSTER AWARDS AND POSTER PITCH

Awards Presenter: **Claas Roehl**, President, NF Kinder – Verein zur Förderung der Neurofibromatoseforschung Österreich
Here, we will discover the winner and runners-up of the ECRD 2024 Poster Awards! We will then continue learning from each other with more pitches from among our top-scoring poster authors. Remember to browse all posters showcased on our interactive online conference platform.

16.15 - 16.45
HIGHLIGHTS HUB

Facilitator: **Geoff Case**, Digital Editor and Senior Associate at RARE Revolution Magazine.
Here is your chance to discover the most important insights and highlights from Day 2 of ECRD. This session will feature short interviews with key speakers and session chairs, summarising the most important discussions and policy outcomes. This interactive catch-up will also provide opportunities for you to share your own perspectives.

CLOSING PLENARY SESSION

16.45 - 17.30

KEYNOTE ADDRESSES

Stella Kyriakides
European Commissioner for Health and Food Safety

Alain Coheur
President, Section for the Single Market, Production and Consumption, European Economic and Social Committee

Jakub Dvoracek
Deputy Minister of Health of Czech Republic

CLOSING STATEMENTS

Avril Daly
President, EURORDIS- Rare Diseases Europe

Virginie Bros-Facer
Chief Executive Officer, EURORDIS - Rare Diseases Europe

DURING THE CLOSING CEREMONY...

Please join us in a Ceremonial signing of the ECRD 2024 Open Letter to European Institutions and country leaders.

Representatives from different stakeholder groups will publicly declare their alignment with the key messages from the rare disease community.

Be part of this significant moment!

REVOLUTIONISING FUNDING STRATEGIES FOR BREAKTHROUGH THERAPIES IN RARER DISEASES

Wednesday, 15 may 2024, 14.00 - 15.30

SESSION DESCRIPTION

This session will explore the need for innovative funding models in the development of therapies for rarer diseases.

We start by acknowledging the limitations of the current therapies' development model, particularly its inadequacy in addressing the unique challenges of very rare diseases. Our discussion will focus on key areas such as:

- Strategies for funding highly individualised clinical trials (n-of-1 and n-of-few) and their effective implementation.
- Exploration of existing, innovative funding models for research and development in small patient populations.
- Discussing the potential of personalised medicine in transforming the treatment landscape for rare diseases.

We will highlight best practices and showcase examples of therapies developed through innovative funding mechanisms and business models. The session will also examine hybrid funding models that bridge the gap between for-profit and non-for-profit approaches, assessing the ecosystem's readiness for such models.

Throughout the session, we will emphasise a collaborative and patient-centric approach, focusing on providing actionable recommendations for rare disease therapeutic development, particularly in rarer diseases. The discussion will centre on fostering patient-focused strategies and collaborative efforts in rare disease research. Click here to listen to the pre-session podcast on the European Rare Diseases Research Alliance

LEARNING OBJECTIVES

At the close of this session, participants are expected to be able to:

1. Summarise the main challenges of funding R&D in rarer diseases.
2. Evaluate alternative business models and funding mechanisms to develop treatments for rare and rarer diseases.
3. Describe the opportunities and challenges of developing clinical research on very few patients.

Session Chair	**Holm Graessner**, Hospital of the University of Tübingen
Speakers	**Robert Bopp**, CFD Foundation eV **Dana Burduja**, Life Sciences and Health, European Investment Bank **Samantha Parker**, IRDiRC and Italfarmaco

Developed with the support of:

Stefano Benvenuti, Fondazione Telethon
Roseline Favresse, EURORDIS-Rare Diseases Europe
Holm Graessner, University Hospital Tübingen
Zoi Kolitsi, I~HD Institute of Innovation through Health Data
Francesc Palau, Hospital Sant Joan de Diu

NO HEALTH WITHOUT MENTAL HEALTH! LET'S CO-CREATE A MENTALLY HEALTHY TOOLKIT

Wednesday, 15 may 2024, 14.00 - 15.30

SESSION DESCRIPTION

The rare disease community has highlighted the lack of holistic support from medical services, where their physical health needs are met but not their psychological and emotional needs. The United Nations recognises this and calls for effective programmes, promoting mental health and psychosocial support, to be implemented through enhanced psychologically informed medical care.

This session will start a co-creation process to develop a new Mentally Healthy Toolkit for the rare disease community, by exploring the existing best practices and tools that can be drawn on and scaled to form the basis for a new Toolkit. As well as providing concrete policy solutions that can support the implementation of the UN commitment into national healthcare systems.

This session will emphasize the vital interconnection between physical and mental health, for people living with a rare condition. We will explore the needs of the rare disease community, at both an individual level, where mental health can be a primary characteristic or a co-morbidity to rare conditions, affecting not just the individual but also the parents, siblings and caregivers; and at a population level, where the rare disease journey impacts directly on the whole community, due to living with uncertainties, medical trauma, isolation, stigma and discrimination.

The session will also draw on and explore existing best practices for psychosocial programmes to test their applicability, transferability and scalability for other rare conditions and communities. Click here to listen to the pre-session podcast episode on **Psychosocial Care: Enhancing Medical Care to be Psychologically Informed.**

LEARNING OBJECTIVES

At the close of this session, participants are expected to be able to:

1. Build the awareness and understanding of the impact of rare diseases on mental health and wellbeing.
2. Leverage the insights gained from existing best practice and tools to alleviate mental health challenges associated with rare conditions for both the individual and the family, to include in EURORDIS' Mentally Healthy Community Toolkit.
3. Identify key recommendations to policy makers and the new European Parliament to develop psychologically informed medical care, that support the UN's call for Mental States to establish psychosocial support for PLWRD and their families.

Session Chair	**Kirsten Johnson**, Fragile X International
Panellists	**Dorica Dan**, Romanian National Alliance for Rare Diseases (RONARD) **Anna Jansen**, University Hospital Antwerp **Lucy McKay**, Medics4RD **André Rietman**, Erasmus University Medical Center **Eva Schoeters**, RaDiOrg – Rare Diseases Belgium **Kym Winter**, Rareminds
Keynote Speaker	**Vinciane Quoidbach**, European Brain Council

Developed with the support of:

Matt Bolz-Johnson, EURORDIS-Rare Diseases Europe
Kirsten Johnson, Fragile X International
Adéla Odrihocká, Rare Diseases Czech Republic
Stefan Živković, National Organization for Rare Diseases of Serbia

THE PATH FORWARD FOR EQUITABLE DIAGNOSIS

Thursday, 16 may 2024, 11.30 - 13.00

SESSION DESCRIPTION

The lengthy and challenging diagnostic process for rare diseases patients, often resulting in misdiagnosis and delayed treatments, remains a pressing issue in Europe. This session echoes the calls to action from Solve-RD, IRDiRC, Rare 2030, and the Czech Presidency, emphasizing the urgent need for a coordinated, European-wide approach to rare disease diagnosis.

Despite significant advances in exome and genome sequencing that facilitated rare disease diagnosis over the last decade, the overall diagnostic rates remain below 50%. Furthermore, the reported average time for accurate diagnosis of a rare disease in 2022 is still about 5 years as highlighted by the 2022 Diagnostic Rare Barometer survey.

Expanding Newborn Screening has emerged as one of the solutions to help shorten time to diagnosis, gaining strong support from individuals living with rare diseases. This session will address patients' experiences on diagnostic odysseys, will look into successful approaches to decrease time to diagnosis by leveraging NBS programmes, through early-genome sequencing in healthcare and also via federated approaches on undiagnosed cases as well as by further disseminating digital tools such as symptom-checkers.

LEARNING OBJECTIVES

At the close of this session, participants are expected to be able to:

1. Explain why the time to accurate diagnosis is still, on average, five years
2. Describe the main current programmes and ongoing activities in NBS
3. Explain how genomic sequencing approaches are used in healthcare to decrease time to diagnosis

Session Chair	**Nick Meade**, Genetic Alliance UK
Speakers	**Simona Bellagambi**, UNIAMO **Jessie Dubief**, EURORDIS-Rare Diseases Europe **Ingo Kurth**, University Hospital Aachen **Graham Shortland**, SWAN (Syndrome Without A Name) Clinic **Miriam Elbracht**, University Hospital Aachen **Petros Tsipouras**, FirstSteps Greece

Developed with the support of:

Gabriella Almberg, UCB
Simona Bellagambi, UNIAMO
Toon Digneffe, Takeda
Roseline Favresse, EURORDIS-Rare Diseases Europe
Kirsten Johnson, Fragile X International
Francesc Palau, Hospital Sant Joan de Diu
Ana Rath, Orphanet

ACHIEVING FULL REACH: OVERCOMING THE LAST CHALLENGES TO ACCESS HIGHLY SPECIALISED CARE

Thursday, 16 may 2024, 11.30 - 13.00

SESSION DESCRIPTION

Healthcare services are best organised as close to the population as possible, where decision makers are best positioned to understand and meet local population needs. This founding principle also holds true for rare diseases but given the small patient population size, healthcare planning for rare diseases is more efficiently organised either at national or pan-regional (European) level where there is a sufficient number of cases to understand the associated needs.

In most European countries, Expert Centres manage a caseload that is big enough to develop the expertise locally and to meet the needs of the patient population affected in their country by the most prevalent rare diseases. However, considering the interplay of factors such as prevalence and incidence rate, expert team availability, and financial implications, it becomes evident that for other less frequent and lower prevalent diseases as well as for rare, complex surgical interventions, even the bigger countries need to arrange cross-border collaboration at a pan-European level or even internationally in order to meet the needs of this patient population.

In this session speakers will discuss the feasibility and opportunities for enhanced pan-European cooperation to plan, fund, contract and organise the delivery of highly specialised healthcare services for highly specialised interventions, under leading Expert Centres that would be connected to national, European and international networks, to ensure safe, accessible and sustainable high-quality care for all. Click here to listen to the pre-session podcast on **Accessing Highly Specialised Care**

LEARNING OBJECTIVES

At the close of this session, participants are expected to be able to:

1. Have a better understanding of the unmet needs and gaps in the provision of cross-border care for certain highly specialised services.
2. Understand the rationale for greater solidarity and collective EU action to ensure timely access to adequate cross-border highly specialised interventions, specifically for rare, complex surgeries.
3. Identify the fundamental components involved in establishing an EU system for commissioning highly specialised services, specifically for rare, complex surgical procedures.

Session Chair	**Enrique Terol,** Permanent Representation of Spain to the EU
Speakers	**Ivo de Blaauw**, Radboud University Medical Centre, Nijmegen **Jan Deprest**, UZ Leuven **Fiona Marley**, NHS England **Miriam Wilms**, Advisory Board of SoMA eV **Nicole Wolf**, Amsterdam University Medical Centre

Developed with the support of:

Matt Bolz-Johnson, EURORDIS-Rare Diseases Europe
Holm Graessner, University Hospital Tübingen
Ines Hernando, EURORDIS-Rare Diseases Europe
Eva Schoeters, RaDiOrg – Rare Diseases Belgium

INNOVATIVE THERAPIES, UNEQUAL ACCESS: BRIDGING THE GAP FOR RARE DISEASE TREATMENTS

Thursday, 16 may 2024, 14.00 - 15.30

SESSION DESCRIPTION

This session will delve into the dynamic landscape of healthcare legislation and access, navigating through key regulatory frameworks and innovative approaches. Our objective is always to advocate for improved legislation for rare diseases, emphasizing a united call to action that aligns with the upcoming Commission plans in 2024. Access to medicines is a multidimensional topic which is influenced by many factors and which encompasses different dimensions.

For many years EURORDIS and other patient advocate groups have been calling for an improved access of orphan medicines towards patients. However, market access is not equal to patient access. The main causes of this situation are related to the high cost of the treatments, the reluctance of payers with regard to the cost-effectiveness of the orphan medicines and the differences in health systems and legislations. We are confronted with the paradox whereby on the one hand more and more treatments for orphan diseases are launched but on the other hand inequalities are increasing and waiting times are growing.

With an important pipeline in front of us based on new technologies such as cell and gene therapies, it is necessary to resolve the paradox. Our objectives for this session include outlining discussions on the implementation of measures, underscoring the need for harmonization, particularly for ultra-rare diseases, and addressing the financial intricacies of launching such measures in multiple Member States in a condensed timeframe.

LEARNING OBJECTIVES

At the close of this session, participants are expected to be able to:

1. Understand and address the challenges that contribute to the widening gap between market access and actual patient access, including high costs, payer reluctance, and disparities in health systems and legislations.
2. Analyze and advocate for improved policies that better support access to orphan medicines for rare disease patients
3. Propose and discuss shared policy measures, especially for ultra-rare diseases, and to explore solutions for the financial and logistical complexities across various Member States.

Session Chair	**Jo de Cock**, Former General Administrator, RIZIV/INAMI
Speakers	**Donatello Crocetta**, UCB **Daniel de Vicente**, Asociación de pacientes ASMD España **Celeste Scotti**, Fondazione Telethon **Christine Leopold**, Utrecht University **Mariangela Pellegrini**, ERN Bloodnet

Developed with the support of:

Gabriella Almberg, UCB
Stefano Benvenuti, Fondazione Telethon
Simone Boselli, EURORDIS-Rare Diseases Europe
Jo de Cock, Advisor to WHO Europe and OECD
Anne-Sophie Lapointe, French Ministry of Health and Solidarity
Eva Schoeters, RaDiOrg – Rare Diseases Belgium

NATIONAL PLANS: EXCHANGING BEST PRACTICES TO FORGE A UNIFIED EUROPEAN RESPONSE TO RARE DISEASE

Thursday, 16 may 2024, 14.00 - 15.30

SESSION DESCRIPTION

This session aims to bring a renewed focus on the national plans and strategies for rare diseases to address the unmet needs of patients and their families.

National plans and national strategies for rare diseases are the common denominator of current public health policy on rare diseases across the EU. The 2009 'Council Recommendation on an action in the field of rare diseases' encouraged most EU Member States and other European countries to adopt a national plan or strategy for rare diseases. While some of them have expired or become obsolete, we observe today a growing momentum, with 13 countries currently having an ongoing plan, and 12 working on designing a new plan, which in some cases is for the first time.

At this crucial juncture, a renewed focus is placed on national plans and strategies to tackle rare diseases. After a bird's eye view of the status quo in Europe, participants will delve into relevant experiences of three countries, whose good practices in designing, monitoring and implementing healthcare measures shall be showcased.

LEARNING OBJECTIVES

At the close of this session, participants are expected to be able to:

1. Learn from each other about the added value of a rare disease national plan or strategy and how adequate national measures can improve care for people living with a rare disease throughout the patient's journey.
2. Refresh how implementing existing EU-wide recommendations are instrumental to build a comprehensive national plan for rare diseases and consider how new or updated European policies and legislations affecting rare diseases can be embedded into national systems.
3. Converge on the added value and the necessity of coordinating national strategies and policies on rare diseases under a singular European framework, with common goals and indicators, thereby streamlining efforts and ensuring consistency in care and support for people with rare diseases throughout Europe.

Session Chair	**Victoria Hedley**, Newcastle University Institute of Translational and Clinical Research
Speakers	**Antoni Montserrat**, ALAN-Maladies Rares, Luxembourg **Francesca Poloni**, Directorate of Health, Luxembourg **Annalisa Scopinaro**, UNIAMO, Italy **Giovanni Paolo Latella**, Directorate of Health Planning, Italy **Jean-Philippe Plançon**, Alliance Maladies Rares, France **Anne-Sophie Lapointe**, Ministry of Health, France

Developed with the support of:

Victoria Hedley, Newcastle University Institute of Translational and Clinical Research
Simona Bellagambi, UNIAMO
Valentina Bottarelli, EURORDIS-Rare Diseases Europe
Ana Rath, Orphanet
Ariane Weinman, EURORDIS-Rare Diseases Europe
Stefan Živković, National Organization for Rare Diseases of Serbia

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Gabor Purman
Kyowa Kirin

Dr Aung Saw
University Hospital of Wales

Olivia Spivack
Erasmus MC

Sara Talarico
ERN-ReCONNET

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ADhoc - An immersive Serious Game that raises awareness among healthcare professionals about announcing a diagnosis of Rare Diseases

Caroline Wernert-Iberg

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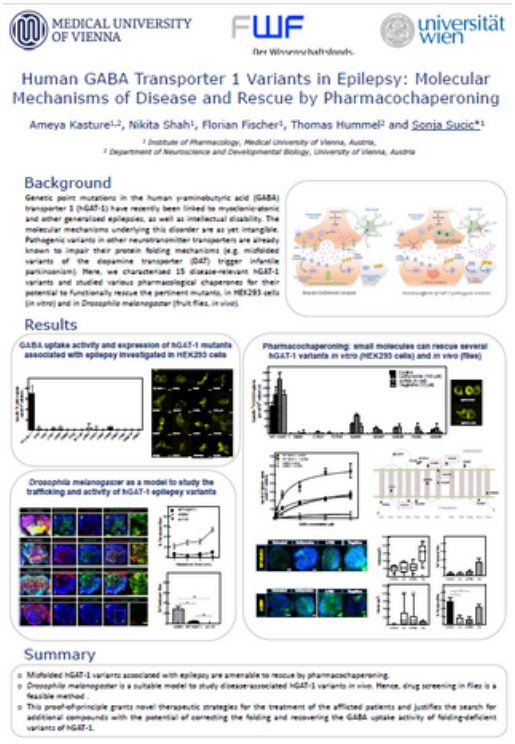
Human GABA transporter 1 variants in epilepsy: molecular mechanisms of disease and rescue by pharmacochaperoning

Sonja Sucic



RUNNER UP

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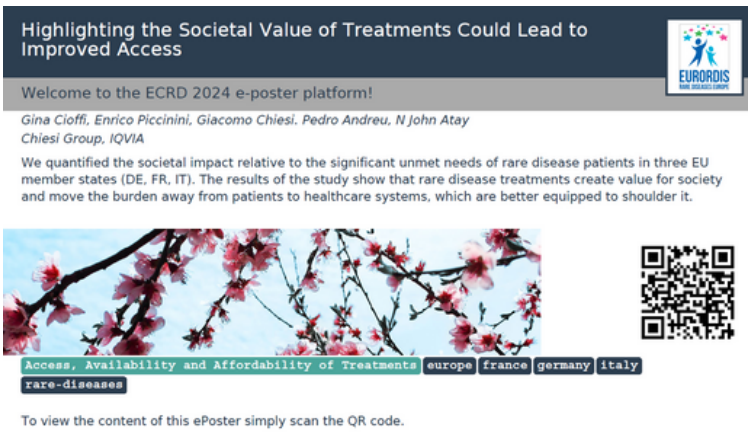
Highlighting the Societal Value of Treatments could Lead to Improved Access

Gina Cioffi



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26 | Ghent University
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94 | International Gaucher Alliance (IGA)
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130 | Orphanet US14-INSERM
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