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

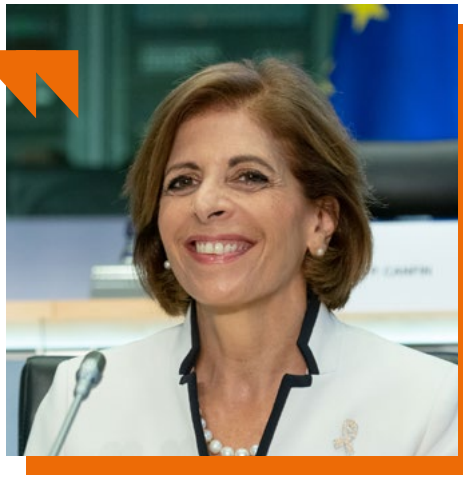
AFMTÉLÉTHON INNOVER POUR GUÉRIR

Co-funded by the Health Programme of the European Union

#ECRD2024

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!

PROGRAMME AT A GLANCE

All times are Central European Summer Time (CEST)

WEDNESDAY
15 MAY, 2024

08.45 – 09.30

ARRIVE EARLY!
Check-in and enjoy a welcome coffee

09.30 – 11.00

DIVE INTO ECRD - EUROPE'S LARGEST, PATIENT-LED, RARE DISEASE POLICY-SHAPING EVENT!

11.00 – 11.30

COMFORT BREAK:
seize the chance to connect and have fun!

11.30 – 12.30

LEARN AND PITCH YOUR IDEAS IN THE POSTER PITCH

12.30 – 14.00

LUNCHTIME!
Join the poster scavenger hunt and organise your own side-meetings!

14.00 – 15.30

REVOLUTIONISING FUNDING STRATEGIES FOR BREAKTHROUGH THERAPIES IN RARER DISEASES

NO HEALTH WITHOUT MENTAL HEALTH!
LET'S CO-CREATE A MENTAL HEALTH AND WELLBEING TOOLKIT

15.30 – 16.10

ANOTHER BREAK FOR SERIOUS FUN!

16.10 – 17.15

JOIN SMALL GROUP DISCUSSIONS

on important topics proposed and selected by you!

17.15 – 18.00

HIGHLIGHTS HUB

18.00 – 19.30

ENJOY MUSIC, FOOD, A 'MYSTERY INGREDIENTS' TASTING COMPETITION AND ENGAGE IN STRUCTURED NETWORKING!

THURSDAY

16 MAY, 2024

08.30 – 09.00

CHECK-IN

grab your welcome coffee, and get ready for action!

09.00 – 09.45

SURPRISE KEYNOTE SPEAKER

Start your day the right way with this interactive catch-up from Day 1 and inspiring keynote speaker!

09.45 – 10.45

POSTER AWARDS AND POSTER PITCH

Continue learning from one another!

11.00 – 11.30

COMFORT BREAK:

connect on a personal level while having fun!

11.30 – 13.00

THE PATH FORWARD FOR EQUITABLE DIAGNOSIS

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

13.00 – 14.00

LUNCH BREAK!

Get ready for serious fun and organise your own side-meetings!

14.00 – 15.30

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:

create memories together!

16.15 – 16.45

HIGHLIGHTS HUB

16.45 – 17.30

BE PART OF THE CLOSING

Full of calls to action, moments of surprise and delight!

- RESEARCH & INNOVATION
- DIAGNOSIS & SCREENING
- ACCESS, AVAILABILITY AND AFFORDABILITY OF TREATMENTS
- MENTAL HEALTH & WELLBEING
- ACCESS TO HIGHLY SPECIALISED CARE
- RARE DISEASE NATIONAL PLANS

IN PARTNERSHIP WITH

Under the auspices of

belgium24.eu

WITH CO-FUNDING FROM

AFMTÉLÉTHON
INNOVER POUR GUERIR

Co-funded by
the Health Programme
of the European Union

IN PARTNERSHIP WITH

efpia
European Federation of Pharmaceutical Industries and Associations

EUCOPE
European Confederation of Pharmaceutical Entrepreneurs AISBL

EuropaBio
The European Association for Bioindustries

ESHG
EUROPEAN SOCIETY OF HUMAN GENETICS

EUROPEAN JOINT PROGRAMME
RARE DISEASES

entidad de utilidad pública
feder
FEDERACIÓN ESPAÑOLA DE ENFERMEDADES RARAS

Huferdis

MedTech Europe
from diagnosis to cure

RaDiOrg
RARE DISEASES BELGIUM

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.

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
Felicitas Riedl, European Investment Bank

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 MENTAL HEALTH AND WELLBEING 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.

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 on the risk and protection factors that influence our 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
Speakers	**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

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
4. Compare a few symptom checkers

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

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.

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 **Ingrid Seinen**, Radboud 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	Coming soon
Speakers	Coming soon

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 **Jean-Philippe Plançon**, Alliance Maladies Rares **Annalisa Scopinaro**, UNIAMO

Developed with the support of:

Gabriella Almberg, UCB
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

HYBRID

ACTION WITHIN REACH

PIONEERING SOLUTIONS FOR RARE DISEASES

HYBRID 15 & 16 MAY 2024

12th European Conference on Rare Diseases and Orphan Products

IN PARTNERSHIP WITH

efpia European Federation of Pharmaceutical Industries and Associations

EUCOPE European Confederation of Pharmaceutical Entrepreneurs AISBL

EuropaBio The European Association for Bioindustries

ESHG European Society of Human Genetics

European Joint Programme Rare Diseases

feder entidad de utilidad pública

Huferdis

MedTech Europe from diagnosis to cure

RaDiOrg Rare Diseases Belgium

Under the auspices of

be EU belgium24.eu

ORGANISED BY

EURORDIS Rare Diseases Europe

orphanet Inserm

EURORDIS
Plateforme Maladies Rares
96, rue Didot 75014 Paris, France
Tel. + 33 (0)1 56 53 52 10
secretariat@rare-diseases.eu

ORPHANET
Plateforme Maladies Rares
96, rue Didot 75014 Paris, France
Tel. +33 (0)1 56 53 81 37
contact.orphanet@inserm.fr