11TH EUROPEAN CONFERENCE ON RARE DISEASES & ORPHAN PRODUCTS

ONLINE

ecrd

27 JUNE ▶ 1 JULY 2022

MISSION IMMUNOPOSSIBLE

PUTTING RARE DISEASE POLICY INTO ACTION

DIGITAL PROGRAMME

AN OFFICIAL EVENT OF THE 2022 FRENCH PRESIDENCY OF THE COUNCIL OF THE EU

FRANCE22
PRÉSIDENCE FRANÇAISE DU CONSEIL DE L'UNION EUROPÉENNE

ORGANISED BY

EURORDIS
RARE DISEASES EUROPE

CO-ORGANISED BY

orphanet
Inserm

WITH THE SUPPORT OF

AFMTÉLÉTHON
INNOVER POUR GUERIR

Co-funded by the Health Programme of the European Union

#ECRD2022

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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.

"Together, we will achieve our commitment to leave no one behind by 2030 so that every EU citizen facing rare diseases can be guaranteed the same opportunities wherever they live in the EU."

OLIVIER VÉRAN, Former Minister for Solidarity and Health, France

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

Quotes from the #30MillionReasons campaign

Because the EU has the science, the healthcare and the industry to help patients suffering from rare diseases. Political will and incentives can turn science into treatments."

Boris, France

So there is no more isolation, no more ignorance about our diseases and to improve our daily lives with appropriate treatments."

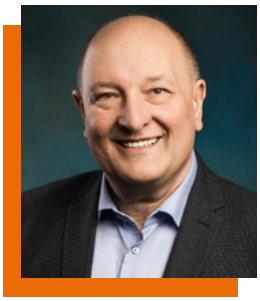
Fani, Greece

Because only Europe can make the invisible, visible, and finally serve all of its citizens needs."

Amanda, Belgium

MESSAGE FROM THE CO-CHAIRS

Ruxandra Draghia-Akli
Global Head, Global Public Health R&D - Janssen Pharmaceutical Companies of Johnson & Johnson



Milan Macek
Head of the Department of Biology and Medical Genetics - National Coordination Centre for Rare Diseases - Charles University, Prague

Ana Rath
Director, Orphanet

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 **11th European Conference on Rare Diseases & Orphan Products 2022** taking place fully online. Globally recognised as the largest, patient-led rare disease policy event, ECRD 2022 has been designated as an official event of the 2022 French Presidency of the Council of the European Union. The theme for this year's conference is **Mission Possible: Putting Rare Disease Policy into Action!**

The COVID-19 pandemic and more recently the war in Ukraine have both exposed the cracks in healthcare systems worldwide and quashed all principles of civil coexistence, international collaboration and respect for human rights and life. For all those whose wellbeing depends on functioning societies, guaranteeing access to care and support, and for those in need of highly specialised medical treatment, as are all people living with a rare disease, the disruptions and interuptions in care and treatment mean further suffering and great risk to life.

It is with this turbulent backdrop that we will embark upon important discussions on how to reach the following 3 visionary goals for people living with a rare disease, inspired by the Rare 2030 Foresight Study and implemented through a new European policy framework for rare diseases.

1. **Ensuring healthy lives and promoting wellbeing for all people living with a rare disease at all ages**
2. **Reducing inequalities for people living with a rare disease**
3. **Building resilient infrastructure, promoting inclusive and sustainable industry and fostering innovation for people living with a rare disease**

These goals are aligned with several of the **UN Sustainable Development Goals (SDGs)** and help accelerate Europe's contribution in achieving them. They also reflect commitments from the current and upcoming trios of EU Presidencies. ECRD 2022 will be the perfect occasion to discuss how to make these commitments and proposals a reality, building upon pivotal European initiatives, for example the European Reference Networks and the European Joint Programme for Rare Diseases that still require a next generation of actions to be brought together under one cohesive strategy.

By maintaining this event online for the second time, we are building upon the success from ECRD 2020, expanding outreach to key stakeholders and building momentum for post-ECRD 2022 implementation while also remaining accessible to all and providing value for money.

New this year! For the first time we are providing our Rare Disease National Alliances with the opportunity to gather key opinion leaders/decision makers on a Member State level to consider how EU policies and national strategies will link in the coming years, with a particular focus this year on how a new policy framework for rare diseases could help play a role. Summaries of these meetings, taking place online during ECRD 2022, will be integrated into the ECRD Executive Summary report, further amplifying the voice from across Europe. Please check out the programme at a glance for full updated information about which National Alliances are participating from which countries.

We are delighted that you are joining ECRD 2022 online 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 50 countries around the world. We value your contribution, your expertise and your opinions. As always, you will have the opportunity to view the high-quality e-posters on display, set up 1:1 calls with your fellow participants and you could even win a prize if you top our interactivity leader board!

Best wishes,

ECRD 2020 Programme Committee Co-Chairs

PROGRAMME AT A GLANCE

ECRD 2022 is taking place fully online and is spread over 5 days.

On the first day of ECRD 2022, the Opening Plenary Session sets the scene for this year's online conference and will be followed by an orientation and networking session along with several parallel Thought Leader sessions. The following three days are dedicated to discussions on how to reach our 3 visionary goals for people living with a rare disease, inspired by the Rare 2030 project and implemented through a new policy framework. A Closing Plenary on Day 5 reminds the audience that rare diseases must be addressed across all of Europe (beyond the EU) and on a global scale. This closing session will also leave participants with a clear call to action in the immediate, medium and long-term future.

All times listed are Central European Summer Time (CEST)

DAY 1
Monday 27June 2022

14:00 - 15:30
Opening & Plenary Session

15:30 - 16:00
Comfort break

16:00 - 17:00
Poster Winners Presentation
Orientation and Networking Session

17:00 - 18:00
Thought-leader Sessions

1. Together4RD – Multistakeholder initiative to unlock ERN collaboration with industry

2. Rare Diseases International – Preparing the foundations of a Global Rare Disease Network

3. Rare Diseases Partnership (Horizon Europe) - Ambition, Vision and Mission

DAY 2
Tuesday 28 June 2022
GOOD HEALTH AND WELL-BEING

11:00 - 13:00
Ukraine session
Mobilising the Rare Disease Movement for Ukraine: EURORDIS and Our Allies' response to the war in Ukraine

14:00 - 15:30
Track A, Session 1
Diagnostic testing technologies, care and treatments: best practices & evidence

Track B, Session 1
Innovative health systems arrangements: best practices & evidence

15:30 - 16:00
Comfort break
Meet the speakers

16:00 - 17:30
Track A, Session 2
Diagnostic testing technologies, care and treatments: best practices & evidence: policy formulation session

Track B, Session 2
Innovative health systems arrangements: policy formulation session

17:30 - 18:00
Meet the speakers

Diagnosis | Healthcare ecosystem | Reducing inequalities | Research & development | Optimal use of data | Availability, accessibility and affordability

PROGRAMME AT A GLANCE

DAY 3
Wednesday 29 June 2022
REDUCING INEQUALITIES

11:00 - 12:00
Language Networking Hub
Connect with fellow participants in your native language

11:00 - 13:00
Swedish Satellite Meeting

12:00 - 13:30
Danish Satellite Meeting (Forberedelsesmøde for patientrepræsentanter **11:00 - 11:45)**

14:00 - 16:00
Track C/D, Session 1
Invisibility as a roadblock towards reducing inequalities

16:00 - 16:30
Comfort break
Meet the speakers

16:30 - 18:00
Track C, Session 2
Inequalities in accessing health and social care

Track D, Session 2
Inequalities in accessing and remaining in education & employment

18:00 - 18:30
Meet the speakers

DAY 4
Thursday 30 June 2022
INDUSTRY, INNOVATION AND INFRASTRUCTURE

10:00 - 12:00
Dutch Satellite Meeting

11:00 - 12:00
Language Networking Hub
Connect with fellow participants in your native language

14:00 - 15:30
Track E, Session 1
Building a seamless health data ecosystem

Track F, Session 1
Making Europe attractive for therapies development

15:30 - 16:00
Comfort break
Meet the speakers

16:00 - 17:30
Track E, Session 2
How to make the best use of ERNs & improve the efficiency of the whole system

Track F, Session 2
Can Europe be attractive and sustainable?

17:30 - 18:00
Meet the speakers

17:00 - 19:30
Romanian Satellite Meeting

DAY 5
Friday 01 July 2022

14:00 - 15:30
Moderated Networking Session

15:30 - 16:00
Comfort break

16:00 - 17:30
Closing Plenary Session

Diagnosis | Healthcare ecosystem | Reducing inequalities | Research & development | Optimal use of data | Availability, accessibility and affordability

A VIRTUAL CONFERENCE

FEATURES OF THE VIRTUAL CONFERENCE

- **Watch live and on-demand sessions** by our over 100 expert speakers until the end of the year
- Meet and exchange with fellow participants during our **moderated networking sessions, open lounges and new language hubs** where you will be able to interact with attendees from your own country! The platform will also recommend **participants with similar interests** as you for **1-1 meetings** or group conversations, helping attendees create connections!
- **Discover more than 230 ECRD e-posters** showcasing the impressive work and research done by patient organisations, healthcare professionals and industry in rare diseases
- Compete with other participants to win a prize on our interactivity **Leaderboard**
- Play with our **Photo booth** and share photos/gifs/boomerangs with your contacts

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 13.5 CPD credits.

CLOSED CAPTIONING & TRANSLATIONS

English closed captioning will be available for all ECRD sessions.

For the opening and closing plenaries (27 June 14:00-15:30 and 1 July 16:00-17:30) it will also be possible to activate closed captioning with French translation.

NETWORKING EVENTS

The online conference platform has been designed with user-friendliness in mind to provide a pleasant 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**.
- Join one of the **Networking Sessions** planned over the week:
  - Mon, 27/06 at 16:00 CET - Poster Winners Presentation, Orientation and Networking Session
  - Wed, 29/06 and Thur, 30/06 at 11:00 CET - Language Networking Hubs: free networking time with virtual rooms divided by language to get to know fellow participants in your native language!
  - Fri, 01/07 at 14:00 CET - Moderated Networking Session
- Pop into the **Open Networking Lounge** to meet more attendees and chat informally. The Lounge is open at the following times (all in CET):
  - Mon, 27/06: 15:30-16:00
  - Tue, 28/06: 11:30-13:00 or 15:30-16:00
  - Wed, 29/06: 12:00-13:00 or 16:00-16:30
  - Thu, 30/06: 12:00-13:00 or 15:30-16:00
  - Fri, 01/07: 11:30-13:00 or 15:30-16:00
- Use the hashtags and post on your favorite social media, you'll appear on the **Social Media Wall** below and in the homepage (#EURORDISAwards2022, #RareDiseaseDay, @eurordis)

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IN PARTNERSHIP WITH

Alliance maladies rares
UNIS POUR VAINCRE

efpia
European Federation of Pharmaceutical Industries and Associations

ESHG
EUROPEAN SOCIETY OF HUMAN GENETICS

EUCOPE
European Confederation of Pharmaceutical Entrepreneurs AISBL

EuropaBio
The European Association for Bioindustries

EUROPEAN JOINT PROGRAMME
RARE DISEASES

entidad de utilidad pública
feder
FEDERACIÓN ESPAÑOLA DE ENFERMEDADES RARAS

MedTech Europe
from diagnosis to cure

NGO COMMITTEE FOR
RARE DISEASES

rare diseases czech republic

RARE DISEASES INTERNATIONAL

SÄLLSYNTA DIAGNOSER
RARE DISEASES SWEDEN

ASSOCIATE PARTNERS

EUROPEAN REFERENCE NETWORKS

EpiCARE

ERKNet
The European Rare Kidney Disease Reference Network

ERN BOND
EUROPEAN REFERENCE NETWORK ON RARE BONE DISEASES

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

European Reference Network
Endo-ERN
European Reference Network on Rare Endocrine Conditions

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

European Reference Network
for rare or low prevalence complex diseases
Network
Adult Cancers (ERN EURACAN)

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)
Coordinator
Assistance Publique - Hôpitaux de Paris, Hôpital Pitié-Salpétrière — France

European Reference Network
for rare or low prevalence complex diseases
Network
Eye Diseases (ERN-EYE)

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

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

European Reference Network
for rare or low prevalence complex diseases
Network
Hereditary Metabolic Disorders (MetabERN)

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

European Reference Network
for rare or low prevalence complex diseases
Network
Connective Tissue and Musculoskeletal Diseases (ERN ReCONNET)

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

European Reference Networks
SKIN

TransplantChild

European Reference Network
VASCERN

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

ITHACA
European Reference Network

ASSOCIATE PARTNERS

ASSOCIATE PARTNERS

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PROGRAMME COMMITTEE **CO-CHAIRS**

Ruxandra Draghia Akli
The Janssen Pharmaceutical Companies of Johnson and Johnson

Milan Macek
Charles University Prague

Ana Rath
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Virginie Hivert
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Rare Diseases Sweden & EURORDIS

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Marcus Guardian
EUnetHTA

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Elizabeth Vroom
World Duchenne Organisation & EURORDIS

Ines Hernando
EURORDIS

Yann Le Cam
EURORDIS

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Thalassaemia International Federation

Dorica Dan
Romanian National Alliance for Rare Diseases & EURORDIS

Fanni-Laura Mäntylä
Young Patient Advocate
City of Vantaa, Finland

Matt Bolz-Johnson
EURORDIS

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Università Cattolica Del Sacro Cuore

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AP-HP Nord Université de Paris

Ariane Weinman
EURORDIS

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Mirjam Mann
Allianz Chronischer Seltener Erkrankungen

Baiba Zieleme
Latvian Alliance for Rare Diseases

Gabor Pogany
Rare Diseases Hungary

Irina Miasnikova
Russian Patients Association

Stefan Živković
Nacionalna organizacija za retke bolesti Srbije

Ulrike Holzer
Pro Rare Austria

Cor Oosterwijk
Patient alliance for rare and genetic diseases Netherlands, VSOP

Miriam Ingram
Genetic Alliance UK

Simona Bellagambi
UNIAMO

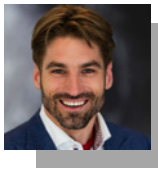
Eva Schoeters
RaDiOrg, Belgium

OPENING PLENARY SESSION

Monday 27th June 2022, 14:00 – 15:30 CET

FACILITATORS

Tamsin Rose



Gerrit Heijkoop

WELCOME AND OPENING REMARKS

Avril Daly, Vice-President, EURORDIS-Rare Diseases Europe

KEYNOTE ADDRESSES

Frédérique Ries, Member of the European Parliament, Belgium

Stella Kyriakides, European Commissioner for Health and Food Safety

REPRESENTATIVES FROM EU COUNCIL PRESIDENCIES

Thomas Lindén, National Board of Health and Welfare, Sweden

Anne-Sophie Lapointe, Head of Rare Diseases Project, French Ministry of Health and Solidarity

Carolina Darias, Minister of Health, Spain

STORIES FROM THE COMMUNITY: THE IMPORTANCE OF A COORDINATED EUROPEAN STRATEGY FOR RARE DISEASES

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

Ana Rath, Director, Orphanet

Stelios Kympouropoulos, Member of the European Parliament, Greece

Irene Norstedt, Director of the People Directorate, DG Research and Innovation, European Commission

CLOSING PLENARY SESSION

Friday 1 July 2022, 16:00 – 17:30 CET

FACILITATORS

Tamsin Rose

Gerrit Heijkoop

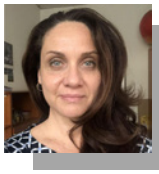
KEYNOTE ADDRESSES

Hans Kluge, Regional Director for Europe, World Health Organisation (WHO)

Jakub Dvořáček, Deputy Minister of Health, Czech Republic

EUROPEAN MEMBER STATE SUPPORT FOR A COORDINATED EU STRATEGY FOR RARE DISEASES

Milan Macek, Institute of Biology and Medical Genetics, Professor of Medical and Molecular Genetics

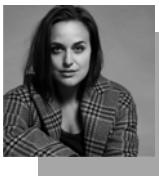


Anna Arellanesová, President of the Board of Directors, Rare Diseases Czech Republic (Ceska Ascociace Pro Vzacna Onemocneni)

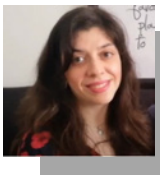
Elvira Martinez, Advocacy & International Relations, FEDER - Spanish Federation of Rare Diseases
(Federación Española De Enfermedades Raras)

Yolanda Agra, Deputy Assistant Director of the Quality Area, Deputy Directorate General of Promotion, Prevention and Quality

PANEL DISCUSSION: YOUNG CITIZEN PERSPECTIVES ON THE IMPACT OF THE RARE 2030 RECOMMENDATIONS



Danielle Drachmann, Executive Director, Ketotic Hypoglycemia International



Oriana de Sousa, Patient Coordinator, Merakoi

Dorothea Dalig, Senior Consultant in the Health Team, Dentons Global Advisors Interel

Adéla Odrihocká, Patient Advocate

CLOSING REMARKS AND THE WAY FORWARD

Yann Le Cam, Chief Executive Officer, EURORDIS – Rare Diseases Europe

THOUGHT LEADER SESSIONS

MONDAY 27TH JUNE 2022- 17:00 – 18:00 CET

Thought Leader Session 1 Together4RD - Multi-stakeholder initiative to unlock ERN collaboration with industry

Chaired by Sheela Upadhyaya, National Institute For Health And Clinical Excellence

Thought Leader Session 2

Rare Diseases International- Preparing the foundations of a Global Rare Disease Network

Chaired by Matt Bolz-Johnson, EURORDIS

Thought Leader Session 3

Rare Diseases (RD) Partnership (Horizon Europe)-Ambition, Vision and Mission

Chaired by Daria Julkowska, European Programme on Rare Diseases

Session Descriptions

Thought Leader Session 1

Together4RD – Multi-stakeholder initiative to unlock ERN collaboration with industry

Chaired by Sheela Upadhyaya, National Institute For Health And Clinical Excellence

27 June 2022 | 17:00-18:00

Together for Rare Diseases is a multi-stakeholder initiative aimed at supporting European Reference Networks (ERNs) to collaborate with stakeholders to pursue opportunities that will address unmet medical needs of people living with rare diseases. The aim is to unlock opportunities for partnerships, particularly with the pharmaceutical industry.

This Thought Leader Session will introduce the work of the multi-stakeholder initiative, Together for Rare Diseases, and offer perspectives from ERN coordinators, patient representatives, industry and policymakers involved in the project on the need to support ERN collaboration with industry in areas that will address the 95% unmet medical needs of people living with rare diseases. The objective of the session will be to outline the steps needed to unlock collaboration and make a call to action for support from the conference attendees.

Agenda: Timing CEST

17:00: Welcome & introduction to Together4RD
Sheela Upadhyaya, Chair of the Together4RD Steering Group

17:15: Keynote and Call to Action
Ondrej Knotek (Renew Europe, Czechia), MEP Champion of Together4RD

17:20: Panel discussion: Unlocking ERN collaboration with the pharmaceutical industry to accelerate research for the 95% unmet medical need in rare diseases

- **Andrzej Rys,** DG SANTE, European Commission
- **Hélène Dollfus,** Chair, ERN-EYE Coordinator
- **Roseline Favresse,** Research Policy & Initiatives Director, EURORDIS
- **Toon Digneffe,** Head Public Affairs & Public Policy - Europe & Canada at Takeda; Member of EUCOPE Executive Board

Discussion moderated by Sheela Upadhyaya, Chair of the Together4RD Steering Group

17:50: Opportunity for audience Q&A
Closing remarks and next steps

18:00: Close

Thought Leader Session 2

RDI – Preparing the foundations of a Global Rare Disease Network

Chaired by Matt Bolz Johnson, EURORDIS

27 June 2022 | 17:00-18:00

Rare Diseases International is collaborating with the World Health Organization to develop a global 'network of networks' that will connect existing collaborations of expert centres and patient organisations. The Global Network for Rare Diseases will support the implementation of Universal Health Coverage, targeting vulnerable and marginalised populations and progressively providing coverage, improving patient access to and coordination of high quality healthcare.

An international Panel of Experts was established in 2019, with leaders from over 100 countries, to codefine healthcare needs, co-design the Network model, and co-create an operational framework for a pilot Global Network for Rare Diseases. Existing clinical networks in rare diseases are leading the development of new models of care for rare diseases. In the EU, the European Reference Networks (ERN) have demonstrated proof of concept for "networked care" that facilitate sharing of experience across the network. National networks have been established in China, France, Japan and now in the USA. Rare Disease "lighthouse" centres that are connected under an existing collaboration or networks will be the flagships for this model of global networked care, and when scaled up and connected under a global network, will create a mature global eco-system to speed up timely diagnosis, generate knowledge, to strengthen health systems locally.

This Thought-Leader Session will explore how healthcare systems in the WHO European region could connect into a Global Network, building on and scaling up the ERN system, exploiting the digitalisation of healthcare and harnessing the collective knowledge from the existing networks to unify an international expert community.

Agenda: Timing CEST

17.00: Introductions & setting the scene
Ines Hernando, EURORDIS

17.05: Concept Model for the Global Network for Rare Diseases
Matt Bolz-Johnson, Rare Diseases International

17.15: EURO Region Readiness Michelle Battye, ERN eUROGEN

17.25 : Patient Perspective Salman Saif, Cure4U (TBC)

17.35 : Panel Discussion & Q&A

17.55: Closing Remarks
Ines Hernando, EURORDIS

Thought Leader Session 3

Rare Diseases (RD) Partnership (Horizon Europe)-Ambition, Vision and Mission

Chaired by Daria Julkowska, European Programme on Rare Diseases

Join us to discover the ambition, vision and mission of the future Rare Diseases (RD) Partnership under the Horizon Europe programme and get answers to your questions!

The aim of the Rare Diseases (RD) Partnership is to improve the health and well-being of 30 million persons living with a rare disease in Europe, by making Europe a world leader in RD research and innovation and delivering concrete health benefits to rare disease patients through better prevention, diagnosis and treatments.

To leave no one behind, RD Partnership will deliver a RD multi-stakeholder ecosystem by supporting robust patient-need-led research, developing new therapies and diagnostic pathways, by utilizing the power of health and research data and spearheading the digital transformational change in RD research and innovation (R&I).

The Partnership will structure the European Research Area (ERA) on RD by supporting the coordination and alignment of national and regional research strategies, including the establishment of public-private collaborations, through research activities all along the R&I value chain, ensuring that the journey from knowledge to patient impact is expedited, thereby optimising EU innovation potential in RD.

Join Daria and other RD leaders to find out more during this interactive session designed for all stakeholders in the rare disease community.

FOCUS ON UKRAINE

Tuesday 28TH June 2022, 11:00 – 13:00 CET

Mobilising the Rare Disease Movement for Ukraine: EURORDIS and Our Allies' response to the war in Ukraine

Chaired by Anastasiia Saliuk, EURORDIS- Rare Diseases Europe

In light of the current humanitarian crisis in Ukraine, the estimated two million Ukrainians living with a rare disease and their families are among the most affected by the conflict, faced with the sizeable challenge of meeting the complex medical needs on their own and falling through the cracks of the humanitarian aid system. In this session, you will hear first-hand from patient organisations in Ukraine and Poland who are responding to the crisis on the ground as well as coordinated efforts by EURORDIS, including the first results of our Rare Barometer survey on Ukraine, and additional initiatives and provisions put in place by European Reference Networks and other humanitarian organisations.

We encourage companies, patient organisations, clinicians and other rare disease stakeholders to join this session to hear about the ongoing needs of Ukranians and the concrete ways you can help.

Speakers:

- **Dr Irina Mykycak,** Deputy Minister of Health, Ukraine
- **Michael Wilbur and Anastasiia Saliuk**, EURORDIS - Rare Diseases Europe
- **Adrian Goretzki,** Healthcare Education Institute
- **Katarzyna Swieczkowska,** Polish Association For Persons with Intellectual Disability
- **Sadie Bynum**, Airbnb.org
- **Dupe Ajayi,** Airbnb.org
- **Helene Dollfus,** Coordinator of the European Reference Network for rare eye diseases ERN-EYE
- **Ruben Diaz Naderi**, SJD Barcelona Children's Hospital, Rare Diseases Virtual Hub**Tetiana Kulesha,** All-Ukrainian Organisation "Orphan Diseases of Ukraine"

SATELLITE MEETINGS

Wednesday 29 June and Thursday 30 June 2022

Swedish Satellite Meeting
Wednesday, 29 June 2022

11:00-13:00 CET

Vi har glädjen att bjuda in dig till Riksförbundet Sällsynta diagnosers digitala valdebatt onsdagen den 29 juni kl 11-13 på zoom.

I höst är det val till riksdag, region och kommun. Inför valet, arrangerar Riksförbundet Sällsynta diagnoser en digital debatt där samtliga riksdagspartier medverkar.

– **Vad säger politikerna?**
– **Vilka partier är redo att ta nästa steg för en nationell handlingsplan?**
– **Hur ser partierna på svaren från Sällsynta diagnosers medlemsundersökning?**

Onsdagen den 29 juni, kl 11-13, ställs de mot väggen.

Passa på och ställ en fråga.
Diagnostik, nya terapier, forskning, regionala skillnader - Vilken är din sällsynta hjärtefråga? Ta chansen och ställ din fråga i chatten under panelsamtalet.
Evenemanget skrivtolkas

Om du har frågor, kontakta **malin.grande@sallsyntadiagnoser.se**

Varmt välkommen!

Danish Satellite Meeting
Wednesday, 29 June 2022
12:00-13:30

Under ECRD kan afholdes nationale satellit-møder mhp at samle sjældne-aktører til status og diskussion af den fremtidige sjældne-indsats. Dette er en invitation til det danske satellit-møde, arrangeret af Sjældne Diagnoser. Mødet er onsdag, d. 29. juni, kl. 12 – 13.30. Kom og vær med! Der er formøde for patientrepræsentanter fra kl. 11, jf. programmet nedenfor.

Baggrund
Corona-tiden har været hård ved sjældne-samarbejdet og der udestår en række opgaver og initiativer: evaluering af den nationale strategi for sjældne sygdomme, forankring af nogle af de initiativer, strategien har resulteret mv. Også European Reference Networks, som har nydt fremme gennem de seneste par år, trænger til opmærksomhed. Samtidig er der aktuelt en række gunstige muligheder som f.eks. det nye Horizon Europe, opfølgning på strategien om personlig medicin, fornyet europæisk fokus på sjældne-samarbejde.

Formålet med satellit-mødet er at samle relevante aktører i Danmark mhp en revitalisering af sjældnearbejdet. Inviterede er følgegruppen til den nationale strategi samt andre relevante sundheds- og socialfaglige aktører, relevante styrelser og ministerier samt patientrepræsentanter. Forud for satellitmødet er patientrepræsentanter velkomne til et forberedelsesmøde, hvor vi sammen ser på baggrunden for ECRD, den nationale strategi og European Reference Networks.

Program

- **Kl. 11.00 – 11.45:** Velkommen til patientrepræsentanterne – gennemgang af baggrund for satellit-mødet v. formand Birthe Byskov Holm og direktør Lene Jensen
- **Kl. 12.00:** Velkommen til – kort intro til det europæiske sjældne-arbejde: ECRD, ny Action Plan for Rare Diseases – Sjældne Diagnoser/EURORDIS (bekræftet)
- **Kl. 12.15:** Arbejdet med den danske strategi – en kort status v. Enhedschef Agnethe Vale Nielsen, Sundhedsstyrelsen (bekræftet) samt efterfølgende input fra andre aktører
- **Kl. 12.45:** European Reference Networks – status v. fuldmægtig Jens Flemming Pedersen, Sundhedsstyrelsen (bekræftet) og udviklingsperspektiver v. vicedirektør Per Jørgensen, Rigshospitalet (bekræftet)
- **Kl. 13.10:** Horizon Europe på Sjældne-området – perspektiver v. fuldmægtig Johan Frederik Mau, Forskningsstyrelsen (bekræftet)
- **Kl. 13.25:** Afslutning: Ses vi igen til efteråret? v. Sjældne Diagnoser (bekræftet)

Romanian Satellite Meeting

Thursday, 30 June 2022
17.00 – 19.30 CET
(18:00 - 20:30 Bucharest Time)

- **Introduction** – Dorica Dan, President, RONARD, Moderator
- **Everyday challenges of a patient with rare diseases** - Patient testimony
- **Priorities of the Ministry of Health** - Alexandru Rafila, Minister of Health
- **Elaborating the National Plan for Rare Diseases and its integration into the National Strategy for Health** – Diana Păun, Romanian Presidency, State Councilor for Health
- **Integrated care services for patients with rare diseases** – Adela Cojan, President, National Health Insurance House (NHIH)
- **International research landscape, reducing inequality within and among countries** - Dr Ruxandra Draghia-Akli, Global Head, Johnson & Johnson Global Public Health R&D

- **Why do we need a European Action Plan for RD?** – Alin Mituta, Romanian MEP

  The impact of a European Action Plan on Cancer - Nicolae Ștefănuță, Romanian MEP
- **Accelerating market access to the therapies that patients with rare diseases and rare cancers need** - Ioana Bianchi, ARPIM
- **Access to therapies and clinical trials for patients with rare diseases** - Răzvan Prisada, ANMDM (TBC)
- **How can Romanians access more programmes dedicated to rare diseases in a way that facilitates cross-border collaboration?** - Emilia Severin, UMF București
- **How can an intersectoral collaboration platform be initiated to develop an integrated care strategy? -** Mihai Tomescu, President ANPDPD / National Authority for People with Disabilities
- **Are rare diseases included in the National Health Strategy?** Adrian Pană, Center for Health Outcomes & Evaluation
- **Why do we need a national register of rare diseases?** Mihai Ioana, UMF Craiova, counselor MoH
- **Do we need a national genetic testing programme in BR and CR? -** Maria Puiu, UMF Timisoara, Vice-President, RONARD
- **The situation of neonatal screening in Romania** - Raluca Teleanu, Hospital Victor Gomoiu Bucharest
- **Ensuring that even the most isolated patients with BR get the services they need** - Lidia Onofrei, MoH, national coordinator of community nursing
- **Opportunities and barriers brought by new technologies (such as genomics) for early detection and screening of rare diseases** - Marius Geantă, Center for Innovation in Medicine
- **Challenges faced by centers of expertise** - Ruxandra Jurcuț, CC Iliescu Bucharest, coordinator CoE
- **Improving access to training for all stakeholders involved in diagnostic and care for RD patients in Romania** - CoE coordinators
- **Questions from participants**

Dutch Satellite Meeting
Thursday, 30 June 2022
10:00 - 12:00 CET

Nederlandse Satellietmeeting: 'Samenwerken in Nederland en Europa'

Op donderdagmorgen 30 juni van 10.00-12.00 uur (inloggen vanaf 9.45 uur) is er een Nederlandstalige satellietmeeting. We bespreken hoe de ERN's beter kunnen worden ingezet voor de Nederlandse zorg aan de hand van de 'Toolkit to support integration of European Reference Networks and national healthcare systems': www.eurordis.org/publication/toolkit-integration-erns.

Verder focussen we op de gewenste samenhang tussen Europese en Nederlandse registers, met het in ontwikkeling zijnde ERN CRANIO register als voorbeeld. Ook staan we stil bij de samenwerking tussen Nederlandse expertisecentra voor zeldzame aandoeningen, zoals dat plaatsvindt via de zogenaamde Connect-websites.

De satellietmeeting wordt georganiseerd door de VSOP en staat ook open voor deelnemers die zich niet hebben geregistreerd voor het ECRD-congres.

GOOD HEALTH AND WELL-BEING

GOAL LEADERS:

Holm Graeßner, ERN RND and University of Tübingen

Victoria Hedley, Rare 2030 and University of Newcastle-upon-Tyne

Elizabeth Vroom, Global Duchenne Foundation and EURORDIS

With the support of:

Matt Bolz-Johnson, EURORDIS

Inés Hernando, EURORDIS

Goal description

The 3rd Sustainable Development Goal to *ensure healthy lives and promote well-being for all people living with a rare disease at all* ages is closely linked to the concept of Universal Health Coverage (UHC). Achieving UHC is about having timely access to diagnosis as well as access to effective, responsive, safe and affordable care and treatments to ensure better health outcomes.

The rare disease community envisions a future where, with increased solidarity, countries will overcome healthcare fragmentation and inequalities in access, through interconnected and comprehensive health & social care systems that answer to the evolving needs faced by the 30 million people living with a rare disease in Europe.

While the Rare2030 Foresight Study provides the overall direction to achieve such a future in the next decade, the rare disease community still needs to define the specific organisational and institutional arrangements that will move us from the aspirational scenario depicted in these recommendations to transforming care delivery and achieving better health outcomes. This goal will on the one hand, identify specific measures to strengthen health systems arrangements to address some of the wellknown access challenges faced by people living with a rare disease. On the other hand, it will address emerging challenges that call for a deeper transformation of our current health systems propose innovative solutions to tackle these.

The sessions under Track A will focus on well-known "access" challenges and explore immediate measure (low hanging fruit) to increase access to effective diagnostic tools, to highly specialised healthcare services and to allied treatments and interventions for all people living with a rare disease across Europe. Whereas Track B will be devoted to imagining new solutions for emerging challenges and to envision pan-European arrangements, under a future European Health Union, for the delivery of highly specialised healthcare for ultra-rare diseases, as well as the delivery of advanced therapy medicinal products (ATMPs).

Each track will feature a workshop showcasing good practices and evidence, followed by a policy formulation session workshop. The workshops and sessions will address jointly upstream (screening and diagnosis) and downstream (healthcare services and treatments) aspects to formulate policy options that consider as much as possible the interdependencies of upstream and downstream associated health services.

Other key components of UHC such as financial protection, well-being promotion and social inclusion will be addressed by the sessions under Goal 2 and Goal 3.

Track A: Strengthening national health systems to improve access to effective diagnostic testing technologies, care and treatments.

Session 1: Tuesday 28 June 2022, 14:00 – 15:30 CET

Good practice and evidence

This session will present evidence and good practice along the continuum of care to inspire short-term policy action at EU and/or national level. Participants will learn what are the main personal and external factors affecting patients' access to diagnosis, according to the findings of the latest Rare Barometer Voices survey on diagnosis. They will also discover how well-designed care pathways, that optimise access to rare disease Expert Centers, may improve patients' experience with care.

Several case studies will showcase how rare disease Expert Centres can arrange care continuity and coordination with social services and will present innovative approaches to improve access to allied therapies for people living with a rare disease.

Chaired by
Elizabeth Vroom, Chair, World Duchenne Organization and EURORDIS Board member

Speakers
Sandra Courbier, Social Research Director - Rare Barometer Programme Lead, EURORDI**S Sanne Bouwman,** Director of international partnerships, ParkinsonNet
Julie Vallortigara, Research Fellow, Department of Clinical and Movement Neurosciences, University College London
Vinciane Quoidbach, European Brain Council
Johanna Blom, Physiotherapist, Neurology Department of the University Hospital of Skåne in Malmö, Sweden
Monika Benson, Executive Director, Dystonia Europe
Eduard Pellicer, Social Worker - Chronic Complex Patients, Hospital Sant Joan de Déu, Barcelona

Session 2: Tuesday 28 June 2022, 16:00 – 17:30 CET

Policy formulation session

This session will build on the experiences and insights shared in Session 1 to propose concrete policy recommendations to improve access to i) effective diagnostic technologies and services, ii) highly specialised healthcare and iii) allied health and therapy services. Speakers will identify specific measures to remove the barriers to access diagnostic technologies for rare diseases and strengthen health systems capacities to treat the increasing number of people that are being diagnosed. The panel will also discuss policy and organisational measures to optimise the design of care pathways to accelerate access to expert care as well as ways to ensure continuous and timely access to allied health and therapy services.

Chaired by
Holm Graessner, Head Research Management Unit, COO Centre for Rare Diseases Tübingen & Coordinator, ERN-RND.
Elizabeth Vroom, Chair World Duchenne Organization and EURORDIS Board member

Speakers
Laurence Faivre, Hospital Practitioner and Professor at the University of Bourgogne, **Hortensia Gimeno,** Associate Director for Research and Clinical Effectiveness, National Health Service of United Kingdom
Holm Graessner, Head Research Management Unit, COO, Centre for Rare Diseases Tübingen & Coordinator, ERN-RND.
Eileen Treacy, Clinical Lead, National Rare Diseases Office

Track B: Inventing innovative health systems arrangements to address new and emerging challenges.

Session 1: Tuesday 28 June 2022, 14:00 – 15:30 CET

Good practices and evidence
This session will showcase a range of good practices in the broad area of 'innovation across the care pathway', starting from accelerated applications of genomic technologies, expansion of newborn screening panels for actionable diseases, and integration of diagnostic approaches for undiagnosed cases, all of which should serve to facilitate an accurate and timely diagnosis and offer the opportunity for all PLWRD to be fast-tracked on an effective pathway.
Larger populations of accurately diagnosed individuals must be accompanied by equally innovative approaches to 'open the door' to curative and transformative therapies; therefore, this session will also consider innovation in health systems to deliver and access highly specialised healthcare services for very rare diseases, intended to speed up the rate at which innovative therapies become accessible to the rare disease community.

Chaired by
Victoria Hedley, Policy Officer, RARE 2030 Foresight Study & University of Newcastle upon Tyne

Speakers
Mark Briggs, Assistant Director of Innovation, Cardiff and Vale University Health Board, United Kingdom
Marie-Christine Ouillade, Board of Directors, AFM-Téléthon & SMA Europe
Tudor Groza, Phenomics Team Lead, European Bioinformatics Institute
Olaf Riess, Head of the Institute of Medical Genetics and Applied Genomics
Francis Pang, Orchard Therapeutics

Session 2: Tuesday 28 June 2022, 16:00 – 17:30 CET

Policy formulation session

'Healthy lives' and greater wellbeing for people living with rare diseases is only possible through a coordinated policy approach which unlocks the potential of innovation in multiple domains, including digital systems, genomic technologies, health systems infrastructure, manufacturing processes, workforce development, knowledge building, and sharing data. Besides spanning multiple disciplines, policies to support innovation in rare disease health systems must often also span many geographical borders.

Building on the good practices and insights shared in Session 1, a panel of experts will discuss policy opportunities to scale-up innovation to ultimately increase life expectancy and improve quality of life for the 30 million Europeans affected by a rare disease or rare cancer (as opposed to the few).

Recognising the need for concerted action for rare disease, the session will discuss some of the core building blocks for a new strategic policy framework for rare diseases, identifying where innovation can be better leveraged to allow EU citizens to benefit from innovative and curative therapies. The pros and cons -and feasibility- of different approaches will be discussed, by considering underdeveloped tools, programmes and approaches which already exist, whilst contemplating what radical future policies might look like.

The session will explore if the existing health legal and institutional framework would allow the flexibility to keep pace of innovation developments as well as fleshing out greater informal or formal collaboration across the European Union, centralising planning and organisations of services, taking a whole-system approach to accelerate adoption and access of innovation into frontline services, access to innovation to all.

Chaired by

Matt Bolz-Johnson, Healthcare Advisor, EURORDIS

Speakers

- **Dirk Vander Mijnsbrugge,** Vice-President Medical Affairs Rare Disease Europe & Developed Asia, Pfizer
- **Nick Meade**, Joint Interim Chief Executive & Policy Director, Genetic Alliance UK
- **Eva Schoeters,** Director, RaDiOrg- Rare Disease Organisation Belgium
- **Edith Gross,** Senior manager, health and social sciences, EURORDIS
- **Wendy van Zelst-Stams**, Clinical Geneticist, Radboud University Medical Centre, Netherlands
- **Victoria Hedley**, Policy Officer, RARE 2030 Foresight Study & University of Newcastle upon Tyne

REDUCED INEQUALITIES

GOAL LEADERS:

Matt Bolz-Johnson, EURORDIS

Ana Rath, Orphanet

With the support of:

Valentina Bottarelli, EURORDIS

Clara Hervas, Edelman

Goal Description

'Systems' are not adapted to the complex needs of people living with a rare disease (PLWRD). Focusing on reducing inequalities for PLWRD involves the identification and the removal of systemic barriers through targeted policy interventions to ensure PLWRD are given the same opportunities. Focusing on reducing inequalities for PLWRD means achieving greater equity, by providing appropriate compensatory policy provisions to eliminate disadvantage and support affected individuals in reaching their maximum level of health and well-being. For this, a holistic approach that looks at the interrelated aspects of physical, mental, and social health and well-being, and that encompasses the entire life-course, is needed.

A major barrier to improving the inclusion and participation of PLWRD and their families in society is the scarcity of knowledge, limited expertise on the field and a lack of awareness of rare diseases and understanding of their impact. As a consequence, this population is psychologically, socially, culturally and economically vulnerable and faces discrimination and specific challenges in health and social care, education, employment and leisure. This, in turn, causes increased impoverishment and isolation. PLWRD can experience this at any point, or throughout, the course of life and additional factors such as gender, living in a rural area or being from a racial or ethnic minority can worsen the challenges. The COVID-19 pandemic has also had a great and disproportionate impact on PLWRD, with already ill-adapted systems being stretched even further. Our most vulnerable in society should be at the heart of any health emergency response, but instead their needs are too often traded-off and left unaddressed, resulting in life-long consequences.

Greater equity for PLWRD is prompted by the removal of these barriers. Equity is founded in cultural and societal values that are essential in order to ensure the rights of PLWRD are upheld. Equity is central to the rights to life, liberty, security of person and cuts across the whole spectrum of the pillars of our society, from education to work, offering the same opportunities for PLWRD to secure an adequate standard of living as well as to enjoy the highest attainable standard of physical and mental health. In addition, focusing on equity for PLWRD can unlock society-wide public good, benefitting not only the millions of PLWRD and their families but also bringing a productive population back into economies as well as crucial improvements in health and social care and social innovation beyond just the rare diseases field.

The complex and holistic needs of PLWRD are gaining in political recognition and support, with the unanimous approval by the United Nations' General Assembly in a new resolution aim at tackling the challenges of people living with a rare disease and their families. The new political endorsement by the UN offers the rare disease community the opportunity to target systemic change and policy development and closing the equity gap between individuals and communities once and for all!
Goal 2 of ECRD 2022 aims to explore the many opportunities to improve the recognition, understanding and knowledge of rare diseases and their impact on the people who live with them,

spearheaded by policy interventions that can address structural barriers in accessing health and social care, and empower self-determination to live life independently through access to education and work.

Track C & D Session 1 sets the framework to navigate the following two topic-based sessions. It considers how rare diseases are invisible in ill-adapted health care and social systems and addresses the preconditions for inclusions and equal opportunities for PLWRD.

Track C Session 2 explores how greater integration of health and social care unlocks better access to health and social care for people living with a rare disease.
Track D Session 2 shows the everyday challenges for a PLWRD to access education and employment while sharing good practices of inclusivity at school and work which trigger greater independence and contribution to society.

Across all Sessions, case studies will be presented whereas panel discussions will formulate policy solutions.

Track C/D Session 1: Wednesday 29 June 2022, 14:00 – 16:00 CET

Invisibility as a roadblock towards reducing inequalities for people living with a rare disease.

People living with rare diseases (PLWRD) are invisible in ill-adapted systems due to the lack of knowledge, recognition and understanding on rare diseases and their impacts on all aspects of life. Inequalities in health-social care, education and work are majorly due to the lack of adaptation to the needs of this population. The greater the complexity, the greater the need for the service-system-society to adapt, and this depends on a combination of awareness, knowledge, willingness as well as tools and practices that empower positive action.

Inequalities experienced by PLWRD can be made visible through disability assessment, at an individual level, and by measuring the burden of rare diseases, at a population level. However, methodologies and practices do not always capture and recognise the complex needs of PLWRD, consequently limiting access to benefits, social protection, and reasonable accommodation.

This session sets the framework to navigate the following two topic-based sessions within Goal 2 of the programme. With the premise that visibility is a pre-condition for inclusion of PLWRD in society, it outlines the evidence and data needed to measure the burden of rare diseases to make them more visible and to inform policy decisions. The session also outlines the infrastructural, technological and policy shifts and tools that support systems to adapt to the needs of PLWRD, and that favour inclusion and equal opportunities in society (policy frameworks like for instance, the European Pillar of Social Rights or the Disability Strategy).

Chaired by:
Ana Rath, Director, Orphanet.

Speakers:

- **Juanita Haagsma,** Assistant Professor, Erasmus Medical Centre Rotterdam Androulla Eleftheriou, Executive Director, Thalassaemia International Federation
- **Flaminia Macchia,** Executive Director, Rare Diseases International
- **Inmaculada Placencia Porrero**, Senior Expert, European Commission - DG Employment, Social Affairs & Inclusion
- **Anne-Sophie Lapointe,** Head of Rare Diseases Project, French Ministry of Health and Solidarity

- **Elvira Martinez,** Advocacy & International Relations, FEDER - Spanish Federation of Rare Diseases (Federación Española De Enfermedades Raras)

Track C Session 2: Wednesday 29 June 2022, 16:30 – 18:00 CET

Inequalities in accessing health and social care for people living with a rare disease.

People living with a rare disease need medical care and follow-up support from different categories of health professionals, often from several different medical specialties, as well as from social workers and other social and local service providers. However, people living with a rare disease face fragmented care pathways and mainstream services that are not flexible enough to take into consideration unprecedented needs.

Health and social care services are not constructed in a way to meet the needs of complex diseases and development disorders. Services have structural barriers to access services leading to vulnerable individuals falling through the gaps in health and social care systems. Complexity of the rare disease means that patient health needs cut across disciplines and there is a lack of multidisciplinarity (or multidisciplinary teams) working (in reality) and a lack of holistic care. Greater integration of health and social care has been shown to directly reduce these inequalities.

Health inequalities show a significant discrepancy across Europe with a dramatic variation in survival rates due to accessing trained, experienced medical team. These inequalities can disproportionately affect rural populations due to limited access to services and further compounded by the lack of willingness to accept a PLWRD in everyday life. Clinical networks and tele-expertise have started to test how to reduce crossborder inequalities.

Chaired by:

Maria Montefusco, Board of Directors, EURORDIS-Rare Diseases Europe

Speakers:

- **Julia Faulkner,** Patient Advocate, Tracheo-Oesophageal Fistula Support (TOFS) UK and EAT
- **Encarna Guillén- Navarro**, Hospital Clinico Universitario Virgen De La Arrixaca, Unit for Medical Genetics
- **Tony Holland,** President, International Prader-Wili Syndrome Organisation (IPWSO)
- **Dorica Dan,** President, Romanian National Alliance for Rare Diseases (RONARD) and Board of Directors member, EURORDIS
- **Isabella Brambilla,** Member and President, Dravet Italia Onlus
- **Wout Feitz,** ERN eUROGEN HCP Network Coordinator
- **Andreas D. Rosenberger,** Centre Leader, National Neuromuscular Centre, Norway

Track D Session 2: Wednesday 29 June 2022, 16:30 – 18:00 CET

Inequalities in accessing education, work and independent living for people living with a rare disease.

To have the same opportunities for economic and social independence, access employment is predicated on acquiring the necessary skills and abilities from school and the education system. Educational systems and schools are not always effective in responding and adapting to the physical and emotional health needs of someone with a complex rare disease, without creating equalities for the individual in accessing the same opportunities in social and educational activities as others. Even with a solid education and having the capacity and desire to work, people living with a rare disease have can face serious stigma and discrimination leading to increased challenges to access employment or returning to work following treatment. This is also the case of their carers who need support to fulfil their caring role whilst remaining in employment.

This session will explore the everyday challenges for a PLWRD to access education and employment and sharing good practice of how schools and business can be inclusive and offer an individual with the same opportunities to contribute to society and be independent. Many PLWRD experiencing the disability gap widening and increased social isolation and stigma in adulthood. The session will also explore societal normal and public perceptions influence inclusion in employment and society, sharing good practice to inform a policy discussion.

Chaired by:
Matt Bolz-Johnson, ERN & Healthcare Advisor, EURORDIS-Rare Diseases Europe.

Speakers:

- **Allison Watson,** Chief Executive Officer and Cofounder, Ring20
- **Oscar Raul Lozano,** University of Valencia
- **Rebecca Scott,** Senior physiotherapist, National Star
- **Jamie Bolling,** Research Developer, Swedish Disability Federation
- **Petia Stratieva,** Retina International
- **Loredana Dicsi**, Membership, Internal Communication & Youth Officer, European Disability Forum

INDUSTRY, INNOVATION AND INFRASTRUCTURE

GOAL LEADERS:

Ruxandra Draghia-Akli, The Janssen Pharmaceutical Companies of Johnson and Johnson

Simone Boselli, EURORDIS

Hans-Georg Eichler, Association of Austrian Social Security Bodies

Virginie Hivert, EURORDIS

With the support of:

Maurizio Scarpa, MetabERN

Anton Ussi, EATRIS

Goal Description

Track E will address the challenge of building a cohesive rare disease health data ecosystem echoing the resilient infrastructure mentioned in SDG 9.

"Within the next 10 years, outcomes, actionable data should be routinely used to accelerate diagnosis, to guide care and management, to support better coordination of care, drug development, HTA and regulatory decision-making and inform health policy."

This vision for a seamless European rare disease health data ecosystem, as described in the Rare2030 Recommendations, envisages a cohesive data landscape where epidemiological, healthcare, research, quality of life and treatment-related data are shared and linked at a European and global level where possible. Sharing of data for different purposes is optimised across infrastructures and countries, relying upon commonly adopted codification systems (Orphanet nomenclature), harmonised standards and interoperability requirements. Under this vision, national data ecosystems are linked seamlessly to the European ecosystem via Findable, Accessible, Interoperable and Reusable (FAIR) data approaches.

Such an ecosystem, centred on robust European Reference Networks (ERNs) and by extension their clinical research capacities, the European Platform on Rare Disease Registration, and other key infrastructures would be well anchored within the European Health Data Space (EHDS), benefitting from this new framework for data sharing, while also contributing to build this space in a way that acknowledges the specificities of rare disease data and is responsive to the needs and expectations of the rare disease community.

So, how can we transform this vision into reality? Is the rare disease community ready to tap into the opportunity that the EHDS represents to address the challenges faced by people living with a rare disease? The answer: not yet.

Using the health data journey as a compass to structure the discussion, these two sessions will help to disentangle the myriad of initiatives and facilitate an honest discussion among stakeholders to identify what data are being collected; when and for what purposes each stakeholder needs to use the data and what would be acceptable conditions to share the health data sets that each is collecting and curating.

How can we make sure that data collected and used upstream (e.g. research/diagnosis/care) can be re-used downstream (e.g. to improve the quality of care/therapeutic development/ regulatory process/policy and HTA decision-making) and vice-versa, and how can this be orchestrated? What are the immediate measures to set the foundations of a cohesive European health data ecosystem for rare diseases that can benefit from and contribute to the larger EHDS?

Track F will address mainly the second part of SDG 9, hence innovation, Europe's attractiveness for investment and medicine development, as well as sustainability. The two main objectives are as follows:

- **How can we keep Europe attractive for development of treatments and interventions for people living with a rare disease?**
- **Can Europe be attractive and sustainable at the same time?**

To approach these questions, we will look at what is needed to fix framework issues if we continue to operate within the current ecosystem for treatment and interventions development and access for patients. What choices could we make if we would like to address the needs of the entire society based on solidarity and equity, as called upon by the Rare 2030 recommendations?

When looking at the medicine lifecycle from development to access, points to be tackled can be easily identified. The first is the so-called 'valley of death' which exists between the basic research and early stage of development, and the late stage of clinical trials. Indeed, this 'valley of death' is not only problematic in the field of rare diseases, but specific issues and ways of de-risking product development in rare diseases are acute and need to be urgently addressed. A major obstacle is linked to the challenges of clinical trials in small patient populations, on a methodological, operational and/ or a financial level. This brings into perspective the importance of the Directive on Patients' Rights in Cross-Border Healthcare (CBHC), the directive for clinical trials and the various funding mechanisms (angel and private investor funds, public, private, and/or public-private partnerships and funding sources). By no means least is the fragmentation of the EU market: products are granted centralized European Marketing Authorisation, but pricing and reimbursement decisions taken at a national or regional level lead to inequalities in access and availability. The current revision of the EU Orphan Medicinal Product (OMP) regulation and of the general pharmaceutical framework is paving the way for some of these issues to be solved by the legislators, but it cannot fix everything. Innovative solutions must also be put forward on the R&D and access fronts in order to unlock the potential of currently disregarded diseases, to ensure the continuity of evidence generation and to allow the defragmentation of the marketplace, i.e. new models of collaboration and funding.

On top of these improvements, we need to reflect on what kind of society we want to live in and how people living with a rare disease want to see the society evolve, while keeping rare disease at the top of the policy agenda. Building on the Rare 2030 recommendations and on the principles of solidarity, equity and social justice, what political choices are needed to combine the EU's attractiveness for companies - ensuring that Europe is a good place for investment and innovation - and the resilience of the healthcare systems for orphan drugs expenditure, reduction of inequalities between countries and within rare diseases? How do we integrate effectively into our society the ground-breaking innovation that tackle the genetic root causes of the great majority of rare diseases?

The central question is: how can we align values across the development chain? From the decision maker's point of view, what criteria should preside within the distribution of resources for healthcare budgets; how do payers become buyers? On the developer's side, where do we stand with regards to ESG (Environmental, Social and Governmental) approaches in view of fulfilling the SDGs? What are the different options to work upstream on assets, fair price, value-based pricing, procurement, sustainable bonds and so on? What would it take to switch from profit-only-based approaches to approaches which bring societal and environmental benefits? Is B-Corp (or Benefit corporations) the way to go for companies? Should there be this type of indicator attached to this value-based decision making on pricing and reimbursement? These are the questions that we would like to discuss with you all!

Track E - The health data challenge: Building a Cohesive Rare Disease Health Data Ecosystem

Session 1: Thursday 30th June 2022, 14:00 – 15:30 CET

Building a seamless health data ecosystem

This session will provide a state-of-the-art of the specific needs of rare diseases within the European Health Data Space; where are we now, where are we going and where do we want to go? How do we connect data to the wider eco-system to allow those who need it to be able to find it and access it when required? This session will explore these specific challenges and opportunities for the rare disease community. How can we orchestrate exchange and what key immediate actions are needed to build a seamless health data ecosystem?

For this, we will get a clear view of the roadmap that is designed in the EHDS (definition, scope, boundaries and timelines) and reflect as well on how it relates to the Members States level. We will then hear from the current pilot that is put in place for rare diseases and see which opportunities it brings for the whole community. Finally, we will get inspired by the COVID situation which seems to show us that the issues are not technical and that when there is willingness and budget, nothing is impossible - as per the subtitle of our conference, mission …possible.

Chaired by
Mark Hanauer, Co-Director and Director of Innovation Strategy, Orphanet

Speakers:

- **Mélodie Bernaux,** Director of Strategy and Transformation, Assistance publique-Hôpitaux de Paris
- **Entela Xoxi,** Senior Scientific Advisor, RWE4Decisions Steering Group
- **Jelena Malinina,** Patient Data Director, EURORDIS- Rare Diseases Europe
- **Jacques van Helden,** Co-director of the Institut Français de Bioinformatique
- **Miroslaw Zvolsky**, Head of the Department of Clinical Classifications at ÚZIS ČR

Session 2 : Thursday 30th June 2022, 16:00 – 17:30 CET

How to make the best use of ERNs & improve the efficiency of the whole system

This session will cover how we generate data, the amount of data generated, for what purposes and for whom. Some key questions are: How can we make better use of data to advance research on rare diseases? How to better enable real-world data (RWD) for legitimate research and decision making? How does GDPR affect research on rare diseases?

In this session, we will try to find answers to these questions while also debunking some myths related to data sharing. Different practical examples and initiatives on health data and real-world evidence will be presented. We will identify the points of discussion, map actions needed and prioritise actions to build from.

The speakers are clinicians/researchers who are working hands-on with data. They will share their experience on how to generate data of good quality, how to organise the data collection better in the context of diagnosis, epidemiology, natural history and more. We also hear what conditions are needed to use or re-use the data, what types of data handling are needed, how the machines use the data (machine learning, AI, natural languages processes, algorithms, keywords). And finally, what does that mean for healthcare professionals, how do we offer tutoring and training for the medical professionals and prepare for the digital doctors of 2030?

Chaired by:
Maurizio Scarpa, Coordinator of the European Reference Network for Hereditary Metabolic Diseases, MetabERN.

Speakers:

- **Ronald Cornet,** Associate Professor, Department of Medical Informatics at Amsterdam Public Health Research Institute
- **Sergi Beltran,** Head of Bioinformatics Unit and Data Analysis Team at Centro Nacional de Análisis Genómico
- **Francesco Cremonesi,** Technical Lead, Datawizard
- **Ronan Fleming,** Associate Professor, National University of Ireland, Galway, & Assistant Professor, Leiden University

Track F: Building resilient infrastructure, promoting inclusive and sustainable industry and fostering innovation for people living with a rare disease

Session 1: Thursday 30th June 2022, 14:00 – 15:30 CET

Making Europe attractive for therapies development for PLWRD- addressing the valley of death

This session will focus on addressing the 'technical efficiency' issues that exist in Europe, focusing specifically on the ongoing revision of the incentives framework, the implementation of the new regulation on HTA cooperation in Europe, and the possibility for more equitable access to authorised therapies provided by new models.
This session will be formatted as a series of three short moderated conversations between speakers to break down the opportunities and challenges laying ahead of us, preceded by an opening introduction by the Chair.

Chaired by:
Ruxandra Draghia Akli, Global Head, Global Public Health R&D at The Janssen Pharmaceutical Companies of Johnson & Johnson

Speakers:

- **Alexander Natz,** Secretary General, European Confederation of Pharmaceutical Entrepreneurs
- **Olga Solomon,** Head of Unit, Medicines: policy, authorisation and monitoring unit, European Commission - DG Health and Food Safety
- **Alicia Granados,** MD. Global Scientific Advocacy Head. Global Medical Affaires Rare Diseases, Sanofi
- **François Houÿez,** Director of Treatment information and Access, EURORDIS
- **Nathalie Moll,** European Federation of Pharmaceutical Industries and Associations, Director General
- **Avril Daly,** EURORDIS Board Member, CEO of Retina International
- **Tamsin Rose**, Senior Fellow for Health, Friends of Europe

Session 2: Thursday 30th June 2022, 16:00 – 17:30 CET

Can Europe be attractive and sustainable at the same time?

This session will focus on addressing the 'allocative efficiency' or on how to distribute resources to rare diseases therapies within budgets. Furthermore, this session will look at the maturing concepts that are gaining momentum to ensure sustainability and affordability in Europe through ethical approaches (ESG indicators, benefit corporation, social pacts, etc.) This session will begin with an introduction to the topic by the chair (Hans Georg Eichler) followed by a series of short presentations to highlight the concepts and progresses made in integrating new ways to address sustainability and accessibility of therapies for rare diseases

Chaired by
Hans-Georg Eichler, Consultant Physician, Association of Austrian Social Insurance Institutions

Speakers:

- **Giacomo Chiesi,** Head of Global Rare Diseases at the Chiesi Group
- **Yann Le Cam,** CEO, EURORDIS
- **Momir Radulovic,** Executive Director, Agency for Medicinal Products and Medical Devices of the Republic of Slovenia
- **Kasha Witkos,** Senior Vice President and Head of International, Alnylam
- **Sarah Garner,** Senior Policy Advisor, Access to Medicines and Health Products, WHO
- **Sarah Emond,** Executive Vice President, Institute for Clinical and Economic Review

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Vertex

POSTERS

1ST PLACE

A Long-term, Prospective, Multicenter, In-clinic and Online Disease Monitoring Program (DMP) for Patients With Long-chain Fatty Acid Oxidation Disorders (LC-FAOD)

Antonio Nino Ramirez[1], Michelle Polowski[1], Kevin McMahon[1], Camille L. Bedrosian[1], Deborah Marsden[1], Monica Yost[1], Kathryn Ray[1], and Jerry Vockley[2]
[1]Ultragenyx Pharmaceutical Inc., Novato, CA, USA; [2]University of Pittsburgh, Pittsburgh, PA, USA

BACKGROUND

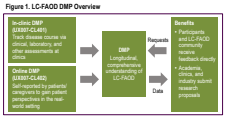
- Long-chain fatty acid oxidation disorders (LC-FAOD) comprise a group of rare, autosomal-recessive disorders of fatty acid metabolism in the mitochondria[1]
- These disorders may manifest in infants, but also commonly present in older children or adults following periods of increased metabolic demand, including physiologic stress, fasting, and prolonged exercise[2]
  - Disease presentation can be heterogeneous, unpredictable, and serious, including life-threatening episodes of rhabdomyolysis, hypoglycemia, and cardiomyopathy, among others[2]
- The addition of LC-FAOD to newborn screening programs began in the mid-1990s
- Recently, triheptanoin, an odd-carbon, medium-chain triglyceride, was approved by the FDA and Health Canada for the treatment of LC-FAOD
  - Given the rarity of LC-FAOD (~1:9,300 worldwide), monitoring the long-term health and outcomes of patients on a large scale is difficult[3]
- Therefore, two studies have been developed as part of the LC-FAOD disease monitoring program (DMP) as an alternative to disease-monitoring surveys and patient registries

OBJECTIVES

- The LC-FAOD DMP is a global, long-term, prospective outcomes study aimed at collecting information from adult and pediatric patients with LC-FAOD for up to 10 years, regardless of disease management or prior participation in triheptanoin clinical trials
- The LC-FAOD DMP includes two distinct, long-term, prospective observational studies designed to provide data from different perspectives (**Figure 1**):
  - Clinic visits (UX007-CL401)
  - At-home data collection (UX007-CL402)
- These studies will assess the long-term safety and effectiveness of treatment in patients with LC-FAOD and establish the natural history, progression, and burden of LC-FAOD to patients and caregivers

OVERALL STUDY DESIGN

Figure 1. LC-FAOD DMP Overview



In-clinic DMP (UX007-CL401)

Overview

- The in-clinic DMP is assessing the long-term safety, including pregnancy, infant, and lactation outcomes, of patients with LC-FAOD
- Secondary objectives include assessments of the long-term effectiveness of triheptanoin treatment on key manifestations of LC-FAOD, including major clinical events of rhabdomyolysis, cardiomyopathy, and hypoglycemia, and the natural history, progression and burden of LC-FAOD, and resource utilization of patients with LC-FAOD
- Patients can either receive or not receive triheptanoin therapy in this observational study and will only have access to triheptanoin through authorized commercial use (if approved in their country) or (if available) an Expanded Access Program, but not from the DMP itself
- Patients will have access to no-cost routine medical follow-up and support for travel to the clinic, as applicable
- Data will be collected and monitored in compliance with Good Clinical Practice (GCP) guidelines for up to 10 years

Study Population

- Approximately 300 participants are planned
- Patients with LC-FAOD who meet the inclusion/exclusion criteria are eligible to participate in UX007-CL401 (**Table 1**)

Table 1. UX007-CL401 Inclusion/Exclusion Criteria

Inclusion Criteria
Males and females of all ages
Confirmed diagnosis of any LC-FAOD subtype. Diagnosis must be confirmed by results of acylcarnitine profiles and/or mutation analysis obtained from medical records or equivalent documentation
Willing and able to comply with all study procedures
Willing and able to provide written informed consent after the nature of the study has been explained and prior to any research-related procedures. For minors in 16 or < 18 years of age, as defined by region: willing and able (if possible) to provide written assent and have a legally authorized representative willing and able to provide written informed consent after the nature of the study has been explained and prior to any research-related procedures
Females of child-bearing potential who become pregnant during the study will be invited to remain in the study. Pregnant females with LC-FAOD will be informed of the study and invited to enroll
Pregnant females carrying affected fetus(es) with confirmed prenatal diagnosis of LC-FAOD will also be informed of the study and invited to enroll
Exclusion Criteria
Presence of a concurrent disease or condition that would interfere with study participation or affect patient's safety in the opinion of the Investigator
Presence or history of any condition that, in the view of the Investigator, places the patient at high risk of not completing the study or would affect the interpretation of study results

- Patients who were previously treated with triheptanoin who enrolled from Study UX007-CL201, UX007-CL202, Investigator Sponsored Trials, or the Expanded-Access Program, as well as adult and pediatric patients who have not previously participated in a triheptanoin clinical trial in LC-FAOD may participate in the DMP
- Participants will be grouped into four cohorts for analysis:
  - Cohort 1: Patients previously treated with triheptanoin in clinical studies
  - Cohort 2: New patients enrolling in the DMP currently being treated with triheptanoin (excluding Cohort 1 patients)
  - Cohort 3: New patients enrolling in the DMP with no exposure to triheptanoin (triheptanoin-naive patients who enroll in the DMP may be on nutrition management alone, medium chain triglyceride supplementation, or other management)
  - Cohort 3a: Patients already enrolled in Cohort 3 of the DMP but who transition to triheptanoin after enrollment

Research Sites

- Approximately 25 research centers will participate in the LC-FAOD DMP, including sites in the United States, Canada, Europe, Latin America, and potentially expanding to other regions (**Figure 2**)

Figure 2. Geographic Distribution of DMP Sites

Data Handling, Record Keeping, and Retention

- Data will be source verified for completeness and accuracy based on standard GCP monitoring methods
  - Validated Electronic Data Capture (EDC) system will be used
  - Monitoring and auditing procedures will be implemented to ensure compliance with International Council for Harmonisation (ICH) GCP standards
  - All records will be retained for at least 25 years after the end of the program or in accordance with national law

Patient Assessments

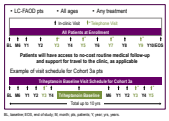
- Age-appropriate assessments (**Table 2**) will be performed in the DMP, depending on the patient's developmental age and physical ability at the time of the visit (**Figure 3**)
- Data for these assessments will be collected only if performed as part of standard of care for in-clinic visits

Table 2. Key UX007-CL401 Assessments

Assessment Type	Measures
Physiologic assessments	• Physical examination • Anthropometric measurements
Rhabdomyolysis assessments	• Serum CK: CK fractions – CK-MM, CK-MB
Cardiomyopathy assessments	• Heart rate • EchocardiogramCardiac biomarkers: Serum troponin (total and TP-1); BNP
Hypoglycemia/liver assessments	• Plasma glucose • Liver enzymes (ALT, AST, GGT) • Bilirubin
Muscle function and motor development	• 12-Minute Walk Test
LC-FAOD MCE/ hospitalization assessments	• LC-FAOD biomarkers and laboratory measures during MCE • Additional disease manifestations during MCE • Nutritional assessments during MCE
Safety assessments	• Pregnancy reporting and outcomes • Neonate and infant outcomes through the first year of life • Lactation reporting and outcomes in nursing mothers and their breast milk-fed infants • Triheptanoin-related SAEs • SAEs and AEs of colon or GI cancer, GI dysplasia, and GI neoplasia • Prior and concomitant medications • Urine pregnancy test (where appropriate)
Survival status	• Patient survival status
QoL assessments	• SF-12, SF-10, or ITQoL • PROs and patient experience • Physician: Clinical Global Impression of Severity
Nutritional management and dosing	• Nutritional assessment of energy and macronutrients • Triheptanoin treatment and compliance • Serum/plasma levels of essential fatty acids and acylcarnitine

AE, adverse event; ALT, alanine aminotransferase; AST, aspartate aminotransferase; BNP, B-type natriuretic peptide; CK, creatine kinase; GGT, gamma-glutamyl transferase; GI, gastrointestinal; ITQoL, Infant Toddler Quality of Life Questionnaire; MCE, major clinical event; PRO, patient-reported outcome; QoL, quality of life; SAE, serious adverse event; SF, Short Form; TP, troponin.

Figure 3. UX007-CL401 Study Schematic

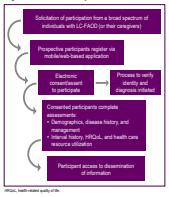


Online DMP (UX007-CL402)

Overview

- The online DMP is being conducted through a mobile/web-based application to longitudinally characterize disease-specific features and obtain a comprehensive understanding of the natural history of LC-FAOD from the patient/caregiver perspective (**Figure 4**)
- Patients can either receive or not receive triheptanoin therapy in this observational study
- This online disease-monitoring program is to be performed in compliance with the protocol, applicable ICH principles, and applicable regulatory requirements

Figure 4. UX007-CL402 Study Schematic



Study Population

- Patients who meet the inclusion/exclusion criteria will be eligible for participation in UX007-CL402 (**Table 3**)

Table 3. UX007-CL402 Eligibility Criteria

Inclusion Criteria
Diagnosis of LC-FAOD, including the following: CPT I or CPT II deficiency, VLCAD deficiency, LCHAD deficiency, TFP deficiency, or CACT deficiency
Willing and able to provide consent or, if a minor, provide assent and informed consent by their legally authorized representative
Access to the internet and an internet-enabled device
Reside in the geographical regions where the platform is supported and approved by applicable IRB/IEC and/or health authority
Exclusion Criteria
Receiving triheptanoin through an Ultragenyx-sponsored interventional clinical trial or Ultragenyx IST

CACT, carnitine-acylcarnitine translocase; CPT, carnitine palmitoyltransferase; IEC, independent ethics committee; IRB, institutional review board; IST, investigator-sponsored trial; LCHAD, long-chain 3-hydroxyacyl-CoA dehydrogenase; TFP, trifunctional protein; VLCAD, very long chain acyl-CoA dehydrogenase.

- Additionally, individuals will be eligible to participate in the online DMP if they:
  - Previously participated in triheptanoin clinical trials
  - Are currently participating in UX007-CL401
  - Are receiving triheptanoin through Expanded Access or Compassionate Use programs
  - Are receiving commercially available triheptanoin via prescription

Solicitation of Participation

- The online LC-FAOD DMP will recruit as many participants as possible, with no prespecified limits
- Information about the online LC-FAOD DMP will be disseminated through clinical, professional, and research groups, and through relevant patient advocacy organizations and support groups
- Patients participating in the UX007-CL401 in-clinic study will be invited to join the online LC-FAOD DMP in geographic regions in which the platform is supported; similarly, patients enrolled in the online DMP will also be permitted to participate in the UX007-CL401 in-clinic study

Data Handling, Record Keeping, and Retention

- Data will be source verified for completeness and accuracy based on standard GCP monitoring methods
  - A validated EDC system will be used
  - All data will be self-reported by the participant (or caregiver reported, when necessary) by responding to a series of questions or surveys
  - All records will be retained for at least 25 years after the end of the program or in accordance with national law

Participant Interactions and Data Entry Process

- Assessments will be completed at scheduled intervals, as summarized in **Table 4**
- History
  - Comprehensive medical history will be collected
  - This section will remain open during the study as a resource for participants
- Enrollment
  - Initial data collection to be completed within 4 weeks
- Longitudinal data collection
  - Repeated measures will be collected at periodic intervals based on the data type and participant's preference
  - Care will be taken to avoid survey and participation fatigue
  - Participants will receive alerts on their mobile device, email, or voicelect message prompting them to complete their assessment within 2 weeks
- Intermittent data collection
  - Additional nonprotocolic data collection will take place based on the data to be collected and the participant's preferences
  - More frequent data collection or intermittent surveys may be given to caregivers of newly diagnosed patients (including newborns) during the first 1 to 2 years of life
  - Participants will have up to 2 weeks to complete these assessments

Table 4. Key UX007-CL402 Assessments

Assessment Type	Measures
Participant registration	• Eligibility questions • Electronic consent
History	• General medical history • Disease-specific history and additional manifestations • Nutritional management and dosing • At-home interventions • MCEs • Health care resource utilization
Enrollment	• Demographics • Disease-specific history and additional manifestations • Nutritional management and dosing • At-home interventions • MCEs • Health care resource utilization • Physical activity • PROs: – SF-12v2 (≥ 18 years old) – EQ-5D-5L (≥ 16 years old) – SF-10 (5 to 17 years old) – ITQoL (2 months to 4 years old) • Participant survey
Longitudinal data collection and intermittent data collection (as requested)	• Disease-specific history and additional manifestations • Nutritional management and dosing • At-home interventions • MCEs • Health care resource utilization • Physical activity • PROs: – SF-12v2 (≥ 18 years old) – EQ-5D-5L (≥ 16 years old) – SF-10 (5 to 17 years old) – ITQoL (2 months to 4 years old) • Participant survey

EQ-5D-5L, EuroQol Five-Dimensions Five-Levels; ITQoL, Infant Toddler Quality of Life Questionnaire; MCE, major clinical event; PRO, patient-reported outcome; SF, Short Form.

SUMMARY

- The LC-FAOD DMP is a multicenter alternative to traditional registries that will be used to consistently monitor disease manifestations and the effect of treatments over a 10-year period
- A unique, core tenet of the DMP is the planned sharing of data with the participants that will include both their own data and deidentified and aggregated data from all participants

DISCLOSURES AND ACKNOWLEDGMENTS

Disclosures: ANR, MP, KM, CLB, DM, MY, KR: Employees and stockholders of Ultragenyx Pharmaceutical Inc. JV: Grant/research support from Reneo Pharmaceuticals and Ultragenyx Pharmaceutical Inc.

Acknowledgments: Jack Rike of Envision Pharma Group provided medical writing support (funded by Ultragenyx Pharmaceutical Inc.), and James Zizzo of Ultragenyx Pharmaceutical Inc. provided editorial support in the preparation of this poster.

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2ND PLACE

Work participation in adults with rare diseases - a systematic scoping review of relevant research.

Thank you much for checking out our e-poster and thank you so much for the opportunity to present our digital poster at this impressive digital Conference
We are three authors that shall present the issue of work participation in adults with rare diseases – a systematic scoping review of relevant research
Brede Dammann we will present information about the background of the study – And, me Gry Velvin will present the scoping review and Trond Haagensen will present - what this work has contributed to.

TRS - national resource centre for rare disorders

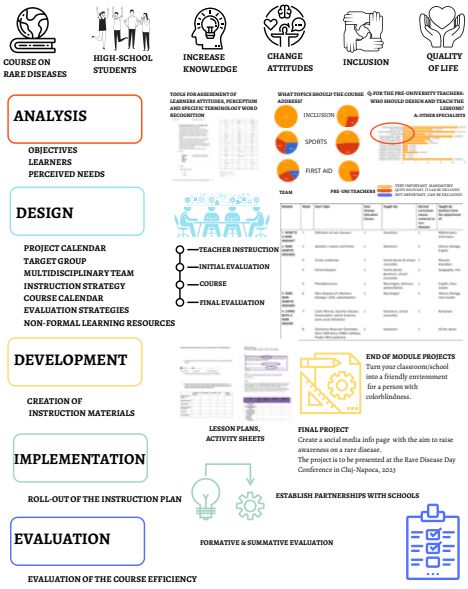
- The Norwegian welfare system is based on the local services.
- Impossible to maintain sufficient knowledge about rare diagnosis in every municipality
- TRS is one out of nine resource centres for rare diseases in Norway. The centres are established to:
  - Build knowledge about the diagnosis
  - Prepare information and cooperate with services that meet people with rare diseases in their daily life.
  - Prepare information about the diagnosis for the people living with rare diagnosis and their families

….and my name is Brede Dammann.
First of all – thank you for the opportunity to address a topic that we think is very instrumental for the wellbeing of people with rare diseases.
In Norway the general welfare services locally based
When it became evident that it is impossible achieve an adequate level of knowledge about rare conditions in every municipality resource centres for rare diseases were established in the 1990. TRS is one of 9 centres that collect and distribute knowledge about the diagnoses.

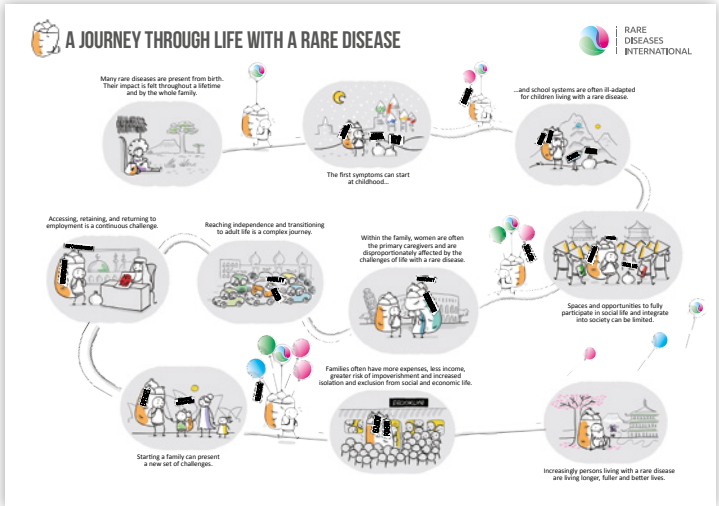
3RD PLACE

USING ADDIE LEARNING MODEL TO DESIGN A MULTIDISCIPLINARY EDUCATIONAL PROGRAM CENTERED ON RARE DISEASES ADDRESSED TO HIGH-SCHOOL STUDENTS IN ROMANIA

Stănculescu I.C, Barabas-Cuzmici Z., Cătana A., Dronca E., Zdrenghea A.C., Mager M.A.,Palade S.



3RD PLACE



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