11TH EUROPEAN CONFERENCE ON RARE DISEASES & ORPHAN PRODUCTS

ONLINE

ecrd

27 JUNE ▶ 1 JULY 2022

MISSION IMPOSSIBLE

PUTTING RARE DISEASE POLICY INTO ACTION

EXECUTIVE **SUMMARY**

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

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Co-funded by the Health Programme of the European Union

#ECRD2022

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KEY FACTS AND FIGURES

61 countries represented

840+ online participants

120+ experts as session chairs, speakers & panelists

4 Ministers and Deputy Ministers of Health

4 Representatives of the European Commission

6 Members of the European Parliament

87 partners

220+ ePosters displayed

4.9 K unique impressions on top LinkedIn post

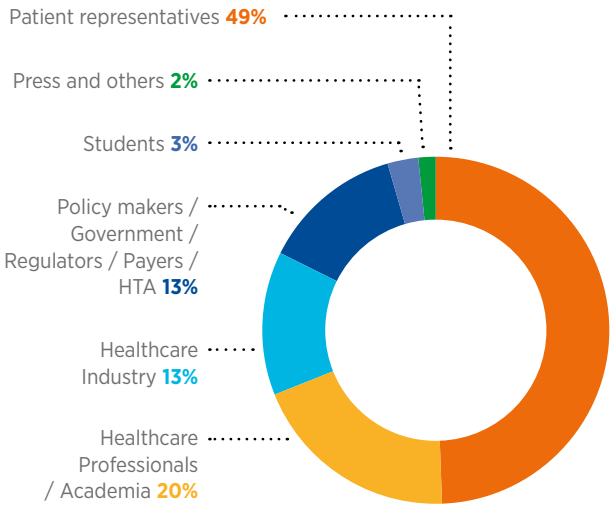
The European Conference on Rare Diseases and Orphan Products is recognised globally as the largest, patient-led rare disease event in which collaborative dialogue, learning, and conversation take place, forming the groundwork to shape future rare disease policies.

The 11th edition of the European Conference on Rare Diseases & Orphan Products (ECRD) was a virtual event for the second consecutive time. By maintaining this event online it has remained accessible to all, reaching over 800 stakeholders of the wider rare disease community from 61 countries.

Over the five days of the conference, participants discussed how rare disease policy should be put into action in Europe to work towards meaningful goals, aligned with the conclusions from the Rare 2030 Foresight Study and the UN Sustainable Development Goals - to improve health and well-being, to reduce inequalities, and to foster innovation. All this has helped to build momentum for post-ECRD 2022 follow-up actions and implementation to secure a better future for all people living with a rare disease.

Through a custom-built platform, registrants were able to participate in 19 sessions with over 120 expert speakers and chairs, and meet with fellow attendees in both facilitated and open networking sessions. The organisers are grateful to the 222 poster authors who contributed such high-quality e-posters.

For the first time ever, four satellite meetings were hosted by Rare Disease National Alliances, gathering key opinion leaders and decision makers on a Member State level to consider how EU policies and national strategies will align in the coming years, with a particular focus on how a new policy framework for rare diseases could drive national efforts.



SETTING THE STAGE FOR ECRD 2022

The progress Europe sees today is a result of policies put in place over a decade ago. Rare diseases have been recognised as a European public health priority - we've seen over 2000 orphan designations, new therapies developed, and the establishment of the 24 European Reference Networks. All this has significantly strengthened the rare disease landscape, but people living with a rare disease in Europe still have vast unmet needs.

Our reality is different now: science has accelerated, technology is embedded in every aspect of our lives and the political ecosystem has changed. It is widely recognised, off the back of the participatory Rare 2030 Foresight Study, that a new rare disease policy framework is needed to set the direction for Europe to leave no one behind.

Significant steps in the right direction that we have seen over the past two years - from the recognition of the challenges faced by the rare disease community globally at the United Nations to the genuine support from the European policy makers - are not to be disregarded. **That is why the theme of ECRD 2022 was designated: Mission Possible, putting rare disease policy into action.**

This ECRD was a critical opportunity for all stakeholders to consider how to transform this exhaustive review of the strategy on rare diseases, as well as the political declarations, into a proposal of concrete actions ultimately creating the ecosystem required to address the unmet needs and persisting inequalities across Europe. This framework would provide an opportunity to set three ambitious goals - aligned with the Sustainable Development Goals - and a number of sub-targets to tackle Europe's challenges:

ENSURING HEALTHY LIVES AND PROMOTING WELL-BEING FOR ALL PEOPLE LIVING WITH A RARE DISEASE AT ALL AGES

REDUCING INEQUALITIES FOR PEOPLE LIVING WITH A RARE DISEASE

BUILDING RESILIENT INFRASTRUCTURE, PROMOTING INCLUSIVE AND SUSTAINABLE INDUSTRY AND FOSTERING INNOVATION FOR PEOPLE LIVING WITH A RARE DISEASE

THE EUROPEAN CONFERENCE ON RARE DISEASES AND ORPHAN PRODUCTS 2022 MIRRORED RECENT AND CURRENT POLITICAL OPPORTUNITIES AND POLICY MILESTONES

- The publication of the Rare 2030 Foresight Study recommendations in February 2021 set the direction for Europe's rare disease policies by 2030 and recommended a new European policy framework on rare diseases.
- The inclusion of rare diseases on the agenda of the EU Trio Presidencies - held by France, the Czech Republic and Sweden. ECRD 2022 was designated an official event of the 2022 French Presidency of the Council of the EU.
- The European Parliament has actively shown its support for an EU Action Plan, through resolutions, debates and open letters.
- The adoption of the Resolution on Addressing the Challenges of Persons Living with a Rare Disease by the United Nations in 2021 reinforced the commitment of the 27 European Union Member States to promote and protect the rights of everyone living with a rare disease and their families.

KEYNOTE SPEECHES

AVRIL DALY
Vice-President, EURORDIS - Rare Diseases Europe

Europe has demonstrated the high added value it can have in protecting & ensuring the health of its citizens. [Rare diseases] must remain a public health priority in the minds of those making the decisions on our future.

FRÉDÉRIQUE RIES
Member of the European Parliament, Belgium

Rare diseases are all but rare, and we need to address it like the health emergency that it is. We need a comprehensive policy framework to connect to all policies and initiatives affecting people living with rare diseases.

STELLA KYRIAKIDES
European Commissioner for Health and Food Safety

Access to orphan medicines differs greatly across Member States. Clearly there is work for us to do here, and that is why the pharmaceutical strategy will address unmet medical needs through a reform of the EU rules on medicines for rare diseases.

THOMAS LINDÉN
Chief Medical Officer Swedish Government

Through national concentration and collaboration, better conditions are created for research, development and knowledge dissemination and the skills of the health workforce are strengthened.

ANNE-SOPHIE LAPOINTE
Head of Rare Diseases Project, French Ministry of Health and Solidarity

The pace of change is uneven across the European continent and, without a general framework at the European level, we risk going backwards, especially in this time of health crisis.

CAROLINA DARIAS
Minister of Health, Spain

[Spain is committed to] the development of a healthcare and research network at a European level that will allow us to achieve the objective of ensuring the best care possible is given to patients living with rare diseases.

KEYNOTE SPEECHES

YANN LE CAM
Chief Executive Officer, EURORDIS - Rare Diseases Europe

Without a coherent strategy, the progress we have achieved in addressing the needs of all people living with a rare disease can be reversed.

ANA RATH
Director, Orphanet

Making it possible to generate and use accurate data on rare diseases is a matter of political will. All starts at the hospital, in outpatient clinics, at the bedside. We have all the recommendations we need, the tools and the models. All is possible.

STELIOS KYMPOUROPOULOS
Member of the European Parliament, Greece

The interconnectedness among the European Regulations is essential. The revision of the OMP regulation cannot be successful without foreseeing for social inclusion. It is not enough for a medicine to be produced and approved, but it should then be accessible and affordable. The necessary social services should be guaranteed even after having taken the medicine.

IRENE NORSTEDT
Director of the People Directorate, DG Research and Innovation, European Commission

The only way we can improve the care for patients with rare diseases is really that we pool the resources and that we work together at an EU level.

HANS KLUGE
Regional Director for Europe, World Health Organisation (WHO)

Now, to drive the implementation of the United Nations Resolution, we need to focus on the 'how', through the development of an action plan for health systems and services to meet the needs of persons living with a rare disease.

JAKUB DVOŘÁČEK
Deputy Minister of Health, Czech Republic

We will continue to support enhanced cooperation and coordination between the Member States and encourage the creation of the European Action Plan on rare diseases.

KEYNOTE SPEECHES

MILAN MACEK
Institute of Biology and Medical Genetics, Professor of Medical and Molecular Genetics, Prague

We have a house which needs to be filled with water, with electricity and all other resources, and we need to improve the access to it. This is where we are, and this is why we need a new policy framework.

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

All in all, we have started a wonderful way towards making a real difference, now we need to keep that momentum.

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

Equity remains a crucial priority for the whole of the community we represent at FEDER.

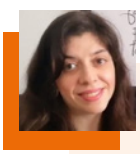
YOLANDA AGRA
Deputy Assistant Director of the Quality Area, Deputy Directorate General of Promotion, Prevention and Quality, Ministry of Health, Spain

Spain is developing a Clinical Patient Management Platform in order to adopt the care model that must be provided by the [European] reference centres.

DANIELLE DRACHMANN
Executive Director, Ketotic Hypoglycemia International

I am not a medical doctor, nor was I educated in how to conduct research – but after years of life-and-death levels of necessary commitment, I consider myself an expert. An expert by necessity....not an expert by choice.

ORIANA DE SOUSA
Patient Coordinator, Merakoi



We cannot work individually. No country has the means nor knowledge to carry all rare diseases. We need a European strategy, we need it so that we are not left behind, but understood and supported.

KEYNOTE SPEECHES

DOROTHEA DALIG
Senior Consultant in the Health Team, Dentons Global Advisors Interel

What we are asking for goes beyond the pharmaceutical intervention and the physiological aspects. We also think that we need better social research to better understand what it means to live with a rare disease.

ADÉLA ODRIHOCKÁ
Patient Advocate

We should not and cannot compromise on the future of our young generation. We need to take action now, to ensure better management of symptoms, improve access to treatments, and guarantee a better quality of life for all.

YANN LE CAM
Chief Executive Officer, EURORDIS – Rare Diseases Europe

The question is no longer IF we need a new European strategy, but HOW and WHEN. We have witnessed the strong political momentum insisting that the current strategy is not enough. This is not the time to relinquish our efforts.

DORICA DAN
RONARD - Romanian National Alliance for Rare Diseases

ECRD 2022 was a great opportunity to link and connect with other people with the same goals and understanding of the reality.

KEY TAKEAWAYS

Over 850 stakeholders (from policy makers to young patient advocates, the pharmaceutical industry, and academia), representing various perspectives and health care-related interests, came together to ask for a new comprehensive plan. The plan includes measurable goals and coordinated actions across a series of many ongoing and a few new flagships.

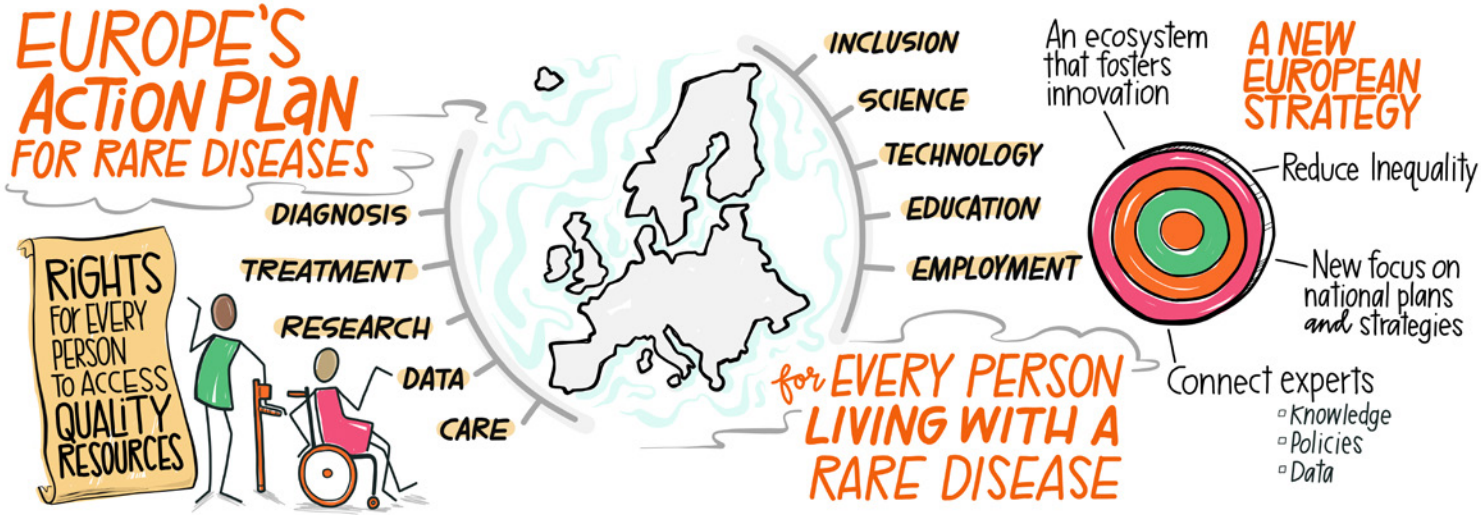
The message was clear: the question is no longer if we need a new European strategy, but how and when. Over the last few years, we have witnessed strong political momentum insisting that the current strategy - over 14 years old - is no longer enough. The Rare 2030 Foresight study highlighted that new, more strategic and coherent policies, under an umbrella strategy, must be established to ensure equal opportunity for people with rare diseases across Europe.

Consequently, 58 partner organisations of ECRD co-signed a letter to call on the EU Commissioner for Health Stella Kyriakides to take forward the conclusions of the conference for a European Action Plan on Rare Diseases.

The common view of the rare disease community is that all legislative and non-legislative actions should be federated under a new Commission Communication and Council Recommendation on rare diseases, revising the framework from 2009. This should see the whole European Union work towards the same common goals, each aligned with a UN Sustainable Development Goal (SDG): promoting health and well-being, reducing inequalities, and fostering rare disease research and innovation.

> **Europe's Action Plan for rare diseases is the necessary step to improve the lives of European citizens living with a rare disease. Success by design would mean linking the needs of a person living with a rare disease from diagnosis, through to data, research, treatment, care and inclusion; enshrining rights to accessing quality care, education and employment; and ensuring that this population can benefit from the cutting edge science and technology that the field is known for. It would strive towards the objectives of both EU4Health and Horizon Europe.**
>
> Extract from letter to Commissioner Kyriakides on behalf of ECRD Partner Organisations, 29 July 2022

From discussions over the five days, the following policy considerations should be taken forward as part of a new, coordinated goal-oriented strategy on rare diseases:



KEY TAKEAWAYS

Enhanced cooperation and coordination between EU Member States

All European countries should work together towards the same targets, to reduce inequalities, improve health and increase innovation.

Connecting the dots between different EU legislative initiatives

The European Union needs a comprehensive policy framework to connect all policies and initiatives affecting people living with a rare disease at the European and national levels, spanning data, research, care, treatment, and social policies. We want to see these actions aligned and developed hand in hand. Efforts at local, regional, national, and European levels should be connected and remain concerted for success.

Updated and renewed national plans for rare diseases

National plans and strategies should provide funds for holistic care to encourage the bridging of health and social care and enable support for care coordinators at centres of expertise.

Rare diseases as a public health priority

The COVID-19 pandemic remains a challenge for the rare disease community and with the war in Ukraine challenges will no doubt increase, shifting the focus of European and national policy makers. Both on a European and national level, we must make rare diseases "commonly known". We must ensure that the needs of this community are not overlooked but well understood, clear and are acted on. Rare diseases must remain a public health priority in the minds of those who make decisions impacting our future.

Public-private partnership

Solutions must be co-designed by the public and private sectors, including civil society and all private actors. There is a need for increased cooperation between national governments, industry, and the patient community to grow awareness and support incentives.

KEY TAKEAWAYS

Policies that work in favour of equity

For people living with a rare disease, equity means social opportunity, non-discrimination in education and work, and equitable access to health, social care, diagnosis, and treatment. We should see more innovative, needs-led, and person-centred policies that eliminate barriers preventing their full participation in society.

Improved data sharing across Europe

Data sharing at the EU level gives the scale needed to advance research, diagnosis, and innovation as well as improve health policy making and direct care. We should focus on robust data exchange in alignment with the European Health Data Space and together with European Reference Networks and Orphanet.

Policies and technologies for accelerated diagnosis

There are vast inequalities in access to diagnosis and the understanding of rare diseases is limited. When a diagnosis is made, healthcare professionals must have the knowledge to provide accurate information on prognosis, adapted care, information, and research. This is one of the main roadblocks preventing access to diagnosis-based, individualised rare disease health care. Awareness-raising efforts amongst frontline healthcare professionals and greater harmonisation across the EU remain central for people living with rare diseases.

Accessible and more holistic assessment of care

By considering the person at the core of all policies, the lines between sectors disappear and the journey of a person living with a rare disease becomes smoother from diagnosis to primary and specialised care, to integration into society, to being not just empowered and engaged but an equal partner with all the other actors in creating treatments and solutions for a better quality of life.

Accelerated innovation development

Europe must strengthen cooperation in research and innovation at all levels: globally as well as between European and national institutions, between the Member States, and between the public and private sectors. There is a need for greater investment in preparing health systems to facilitate access to ground-breaking innovations and technology for people living with a rare disease. Collaboration is needed to transform Europe into an environment where research and development in rare diseases can thrive at scale. This would support the development of formalised multinational and multistakeholder partnerships to harness EU expertise and create an ecosystem that encourages the development of innovative therapies, particularly for underserved rare diseases.

CONFERENCE GOALS

GOAL 1

GOOD HEALTH AND WELL-BEING

GOAL 2

REDUCED INEQUALITIES

GOAL 3

INDUSTRY, INNOVATION AND INFRASTRUCTURE

GOOD HEALTH AND WELL-BEING

GOAL LEADERS:

Holm Graeßner, ERN NMD 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

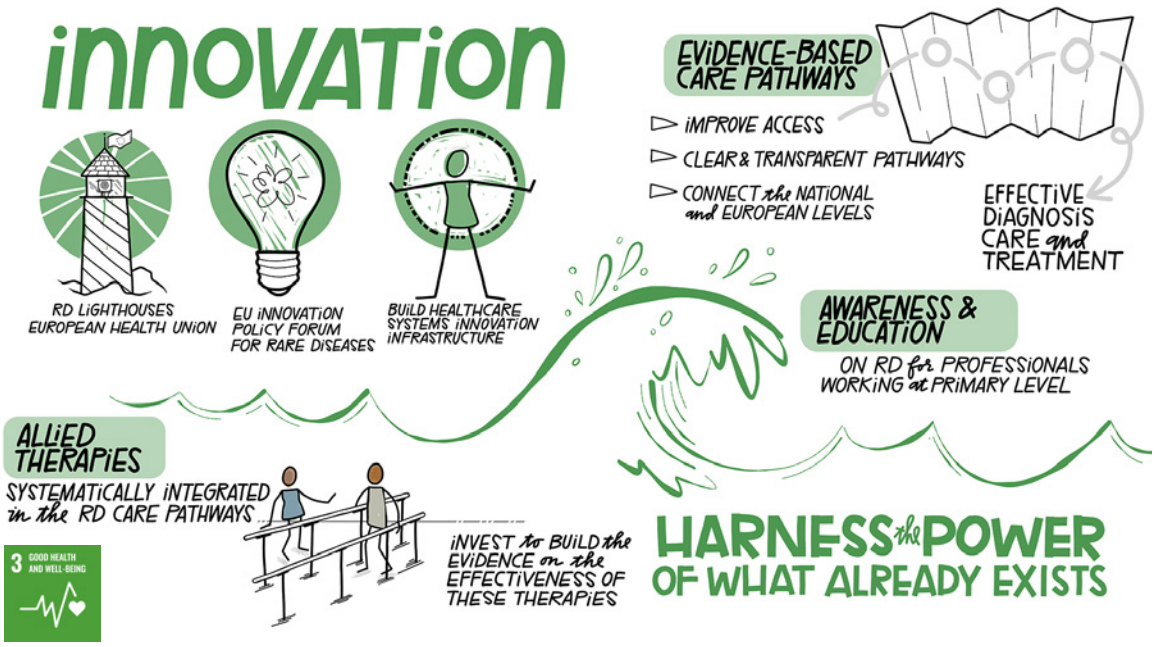
Sessions under Goal 1 explored actions to be taken to work towards achieving SDG 3: Ensuring healthy lives and promoting well-being for all people living with a rare disease at all ages.

QUESTION 1: HOW SHOULD WE STRENGTHEN NATIONAL HEALTH SYSTEMS TO IMPROVE ACCESS TO EFFECTIVE DIAGNOSIS, CARE AND TREATMENTS?

The average time to diagnosis has not changed much in the last decade, receiving a referral to a centre of expertise is down to luck, and allied therapies, which are key drivers in a good quality of life, are not being systematically integrated in rare disease care pathways. With clear mechanisms and structures to improve quality of life and adding years to life, this is an unacceptable loss of opportunities in better harnessing what already exists!

To improve access to timely diagnosis, ensure inclusive, timely and affordable access to high quality care, participants in the sessions agreed that policy action is needed to:

- Raise awareness and education on rare diseases for primary healthcare professionals, for example use the European Reference Network training academy to target GPs, so that they know where to find reliable sources of information.
- Establish clear rare disease care pathways to access Centres of Expertise at the national level, and at EU level where needed.
- Build the evidence base on the effectiveness of allied therapies (speech, occupational and physical therapies) to systematically include these therapies in the rare disease care pathways, as this can optimise the child's development trajectory.
- Organise care for rare diseases based on the size of the eligible population. It should be large enough to understand and meet their needs. In particular, this is needed to develop an EU-wide end to end approach for joint funding, planning, commissioning, and contracting from designated European Centres of Expertise (or, "European Rare Disease Lighthouses"), to cater healthcare services either for very low prevalent diseases (<500 cases EU-wide or <250 cases at a national level) or for highly complex OR for complex procedures where the number of interventions at national level is too low to develop and maintain the competence and skills required.



QUESTION 2: HOW CAN WE EMBRACE INNOVATION TO ADDRESS NEW AND EMERGING CHALLENGES FOR RARE DISEASES?

Innovation is at the heart of rare diseases. No one stakeholder or institution alone is responsible nor can implement innovative practices: to get it right requires a partnership approach. Massive challenges still exist at the national level and EU level in terms of fragmentation in the organisation and delivery of innovations, creating stark inequalities (between diseases and within communities) in terms of access to innovation from basic research through to diagnosis and care delivery.

We still lack a formal forum to build consensus and discuss innovation policies, hence IMI, IHI and Horizon Europe projects are often used to discuss and advance policy options. Is this the right setting for policy development where policy makers are largely absent?

To successfully embrace innovation across all rare diseases, participants in the sessions agreed that policy action is needed to:

- Create an EU innovation policy forum to share and learn together, anchored with well functioning national plans/policy frameworks (steered by multi-stakeholder policy groups). Innovation is rapidly developing, and policy should steer this, but it keeps falling behind scientific advancements. This is a fast- moving field so we need to have nimble policy-making. An EU innovation policy forum would facilitate early dialogues, allow Member States to share and replicate good practice, shining a light on investment into innovation related policy development to increase the ability of systems to adopt new innovations. Participants developed the concept: 'innovation ready countries'.
- Drive innovation partnerships. Better alignment and connection between different networks is required to have a systems approach to innovation. This should include clinical networks (ERNs & National Networks), technological networks, analytical networks and supply networks.
- Put a strategic focus on building healthcare systems' innovation infrastructure. Innovation centres should be connected through a national infrastructure and platform (e.g.: Advanced Therapy Medicinal Products - ATMPs). There is a need for the development of institutional readiness toolkits and for specific guidance on cross-border healthcare to support access for treatment abroad (e.g.: ATMP)
- Expand pockets of innovations and bring in new diseases into Newborn Screening panels in new countries. Participants agreed that the concept of actionable is very new for all!
- Ensure public and patient dialogue to inform and educate on innovation development. EURORDIS plays a significant role in continuing to support National Rare Disease Alliances. We need to continue informing and educating organisations on innovation to support their engagement on this field.

REDUCED INEQUALITIES

GOAL LEADERS:

Matt Bolz-Johnson, EURORDIS

Ana Rath, Orphanet

With the support of:

Valentina Bottarelli, EURORDIS

Clara Hervas, Edelman

Sessions under Goal 2 explored actions to be taken to work towards achieving SDG 10: Reducing inequality within and among countries (as well as SG1, SG3, SG4, SG5, SG8 & SG9)

QUESTION 1: HOW CAN WE MAKE RARE DISEASES 'VISIBLE' SO THAT HEALTH AND SOCIAL CARE, EDUCATION AND EMPLOYMENT SYSTEMS ARE BETTER ABLE TO ADDRESS THEIR NEEDS, AND SUPPORT GREATER INCLUSION AND EQUAL OPPORTUNITIES FOR PEOPLE LIVING WITH A RARE DISEASE IN ALL ASPECTS OF SOCIETY?

The invisibility of the needs of people living with a rare disease is a key driver in the absence of political commitment to address inequalities. Patient groups play a fundamental role in making the needs and inequalities of their community visible but it takes time to mature advocacy based on population data to focus policy actions. During the session, the European Commission invited people living with a rare disease to join fora for discussions to better understand their experience and make their disabilities visible.

The new UN Resolution on addressing the challenges of persons living with a rare disease and their families is a major step towards reduced inequalities, shining a political spotlight on the inequalities people living with a rare disease face.

"The recent crises have unveiled the fragility of the system in maintaining equality and in protecting the most vulnerable." Ana Rath, Orphanet.

"The VISIBILITY of the rare disease population's needs is the prerequisite to reduce inequalities globally." Flaminia Macchia, Rare Diseases International

"When it can't be cured it can be cared. We need data on disability, it contributes to invisibility and is a barrier. We need data to understand a person living with a rare disease full set of needs. Without data, we cannot move forward." Ana Rath, Orphanet

To improve the visibility of rare diseases across every area of life, to better address their needs, participants in the session agreed that policy action is needed to:

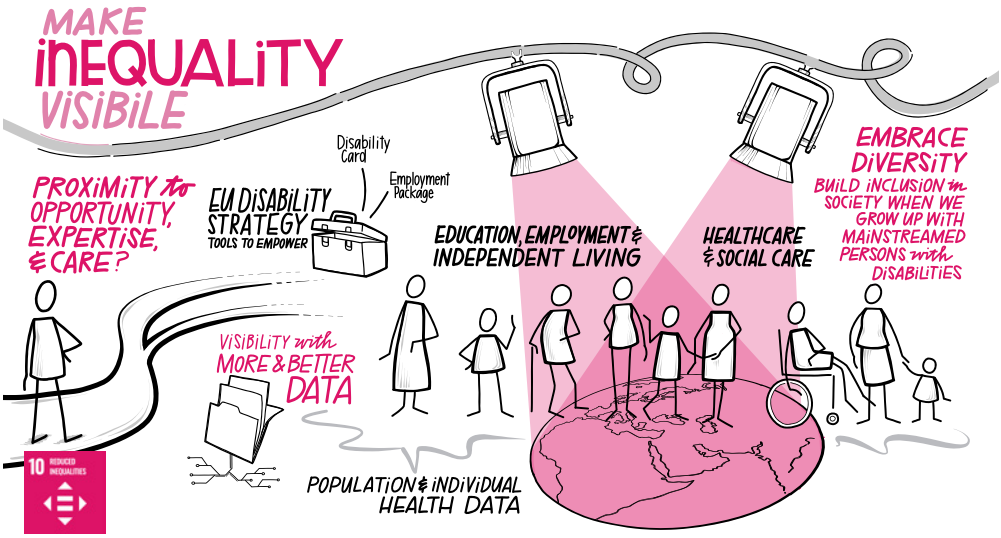
- Establish common goals that are specific to rare diseases - so that regional and national authorities follow the international legitimacy to drive local change.
- Pool data to make sure the needs of a person living with a rare disease are visible on a population level.
- Improve the burden of disease data. Currently rare diseases are lumped together or hidden in bigger categories. Incentives (funding) is needed to collect burden of illness studies and data to make visible the unique needs and inequalities people living with a rare disease face at a personal and population level.
- Create consultations and other opportunities for feedback to maintain this constant dialogue.

QUESTION 2: HOW DOES A GREATER INTEGRATION OF HEALTH AND SOCIAL CARE UNLOCK BETTER ACCESS TO HEALTH AND SOCIAL CARE FOR PEOPLE LIVING WITH A RARE DISEASE?

According to the European Commission's Disability Strategy, 52% people living with a disability feel they are discriminated against. The pandemic has also delayed the implementation of integrated care policies. The main obstacle in defining policy solutions is how to define 'disability', which makes disability assessment and a common disability card hard to implement.

Significant progress was shared that has been made in case management/rare diseases training in mainstream / community services e.g. ECHO training for community nurses, however much remains to be done in holistic care "through life" i.e. beyond paediatric age.

The WHO's five strategies on person-centred integrated care should be the compass for any local actions in this field, and the opportunity of a new EU strategy on rare diseases offers a chance to strengthen actions in this area.



To ensure a greater integration of health and social care for people living with a rare disease, participants in the session agreed that policy action is needed to:

- Renew the partnership approach at a national level to renew implementation of this policy. Integrated care remains valid and relevant to address the needs of people living with a rare disease.
- Ensure the disability assessment as an entry point for access to social support in most welfare systems. The lack of information on the disabilities associated with rare diseases generates a negative impact on social care for persons living with a rare disease.
- Follow up on the EU Disability Strategy including, the European Disability Card which ensures that the mutual recognition of the disability status across the EU will also allow access to social security benefits, not only to internal market services. This would make a difference to both people living with a rare disease and living with disability.
- Turn best practices in the field of integrated care into policies and programmes, which are financially supported, developed and implemented collectively and monitored in collaboration with key stakeholders including patients and families.
- Consider holistic care "through life".

QUESTION 3: HOW CAN PEOPLE LIVING WITH A RARE DISEASE HAVE THE SAME OPPORTUNITIES FOR ECONOMIC AND SOCIAL INDEPENDENCE, THE SAME ACCESS TO EDUCATION AND EMPLOYMENT, WITHOUT THE TRADE OFF BETWEEN THE HEALTH AND SAFETY OF AN INDIVIDUAL, WITH A LOSS OF BASIC RIGHTS TO EDUCATION, SOCIAL INCLUSION AND PARTICIPATION?

"Education creates the foundations for combatting poverty and for creating fully inclusive societies"..."Inclusive education is not a favour but an obligation In the Convention on the rights of people living with disability"..."Right to self-determination, reasonable accommodation, and access to quality jobs and fair wage, is not about what support/ adjustment employers are willing to give, but rather what the employees need." Loredana Dicsi, European Disability Forum.

Progress has been made in advocacy to address health and social inequalities, but there is much more to be done in education, employment, and independent living.

Defending rights for inclusive education is important in creating an inclusive society: specialist education at a specific moment can support a young person developing self-esteem, and acquire skills and autonomy needed in independent living. However, to get there, training and support for teachers and employers in rare diseases is needed.

Digital inequalities were highlighted, as people with a disability have less access to internet and digital technologies, which entrenches digital inequalities throughout school and employment.

Many people living with a rare disease experience the disability gap widening and increased social isolation and stigma in adulthood. This is particularly seen in employment.

To ensure the same opportunities for economic and social independence for people living with a rare disease, participants in the session agreed that action is needed to:

- Train and support teachers and employers in the needs of people living with a rare disease.
- Engrain the responsibility of both the employer, to make reasonable adjustments, and the employee, to continue to be trained and develop for people living with a rare diseases, as a significant lack of skills and knowledge among employers remains.
- Alongside EDF, to set common standards for what "reasonable" means (reasonable accommodation, etc.)
- Follow the launch of the "employment package" when launched in September, as part of the EU Disability Strategy. For example allowing the accumulation of benefits when entering into employment.

INDUSTRY, INNOVATION AND INFRASTRUCTURE

GOAL LEADERS:

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

Simone Boselli, EURORDIS

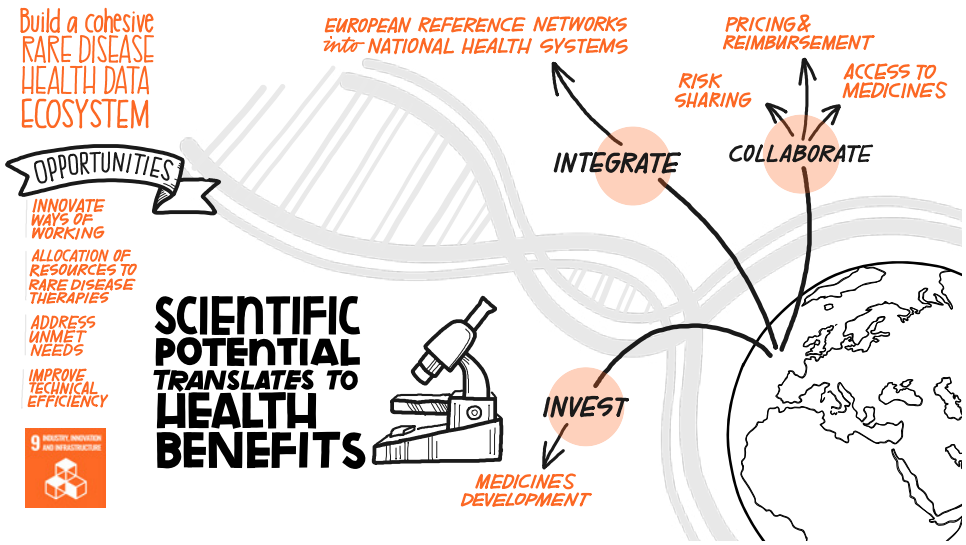
Hans-Georg Eichler, Association of Austrian Social Security Bodies

Virginie Hivert, EURORDIS

With the support of:

Maurizio Scarpa, MetabERN

Anton Ussi, EATRIS



Sessions under Goal 3 explored actions to be taken to work towards achieving SDG 9: Building resilient infrastructure, promoting inclusive and sustainable industry, and fostering innovation for people living with a rare disease (as well as SDG 3 and 10)

HOW CAN WE ENSURE SCIENTIFIC POTENTIAL IS TRANSLATED INTO ACTUAL HEALTH BENEFITS FOR PEOPLE LIVING WITH A RARE DISEASE?

This session addressed the challenges of building a cohesive rare disease health data ecosystem as well as ensuring Europe's attractiveness for investment and medicine development and sustainability.

Science offers an unprecedented opportunity to address the unmet needs of people living with a rare disease. To ensure this potential is translated into actual health benefits for our community, we look on one hand on how we can best distribute resources to rare disease therapies (allocative efficiency), and on the other at how to improve current processes (technical efficiency).

There are innovative ways to work together as a community to bring innovation to rare disease patients in a sustainable manner, for example:

- By having a more collaborative approach to pricing & reimbursement and accelerating / facilitating the conclusion of risk-sharing agreements
- By reimagining access to medicines through the development of Public Private Partnerships

If we -as the patient community- trust that society will bring to us better health, we are ready to give society our data in return to foster research and provide evidence to inform decisions, so we can build cohesive rare disease health data.

In order to do that, participants in the session agreed that policy action is needed:

- European Reference Networks that are fully integrated into national healthcare systems
- European Reference Networks that are enabled to provide data to federated data platforms in a FAIR way and link that back to clinical care, and provide knowledge to structure data

This recalls that a holistic overarching approach is indispensable to have an impact on the lives of people living with a rare disease, in line with the overarching goals of this conference. This makes sense only in the framework of a coordinated, integrated, comprehensive EU strategy that would increase the impact of services in support of diagnosis, prognosis treatment and stratification.

SATELLITE MEETINGS

On 29 and 30 June, four Satellite Meetings took place as part of ECRD 2022, hosted by the National Rare Disease Alliances of Sweden, Romania, Denmark and The Netherlands. Each provided an opportunity for representatives from national authorities, professional organisations, industry and rare disease experts to come together to discuss how an EU Action Plan for Rare Diseases would support their objectives on a national level. With each country having its own challenges and priorities, the Satellite Meetings allowed experts to reflect on the general messages of the conference within their own national context.

ROMANIAN SATELLITE MEETING

61 participants joined the Satellite Meeting hosted online by the Romanian National Alliance for Rare Diseases, around a wide range of topics. Speakers included the Romanian Minister of Health, Dr Alexandru Rafila, the State Counselor at the Romanian Presidency, Mrs Diana Loreta Paun, Members of the European Parliament, academia, and patient advocates.

The debate focused on a European Action Plan on Rare Diseases, research, European Reference Networks, and intergrated care, amongst other issues. The debate identified the main challenge in the Romanian context as being around funding: Limited budgets do not match the vision for treating patients.

The overall conclusions of the meeting set out a series of steps for Romania:

- The need for a national rare disease registry for rare diseases;
- The need to extend the number of rare diseases in Newborn Screening;
- Increased training for family doctors and community nurses on case management of rare diseases, including connecting them to the Centres of Expertise;
- Accreditation of more Centres of Expertise and creation of a national rare disease network;
- Integration of the National Plan on Rare Diseases in the wider National Health Strategy, with adequate budget allocation.

It was recognised that bringing stakeholders together at the same table was a crucial step to finding areas of collaboration, especially in the identified next steps of contributing to the National Health Strategy debates, and promoting the needs of people living with a rare disease at national and international levels.

SATELLITE MEETINGS

DUTCH SATELLITE MEETING

The Dutch Satellite Meeting, organised by VSOP, the national patient alliance for rare and genetic diseases, and chaired by Dr Cor Oosterwijk, Director of VSOP, saw three presentations looking at the interaction between European Reference Networks, Dutch patient organisations, and Centres of Expertise.

The first presentation by Mariette Driessens, PhD, policy officer at VSOP, focused on the VSOP project aimed to develop a digital platform to connect all relevant Dutch patient organisations with their related ERN and ePAG. During her speech, Ms Driessens also expanded on the importance of the EURORDIS Toolkit to support the integration of European Reference Networks and national healthcare systems.

The debate was followed by Daphne Stemkens, MD, another policy officer at VSOP, who presented the websites that VSOP develops for Networks of Expertise: cooperating Centres of Expertise for a single rare disease that are willing to present their medical services and research on a joint website for both patients and medical professionals outside the centres. It was noted that the Networks of Expertise that present their activities on a joint website are successful instruments for helping patients and caregivers look for the right medical care and/or clinical research. She noted, however, that more cooperation is needed between, and amongst, patient organisations, national networks of Centres of Expertise and European Reference Networks.

Prof Irene Mathijssen, a surgeon at Erasmus Medical Centre Rotterdam and coordinator of ERN CRANIO (for rare and/or complex craniofacial anomalies and ear, nose and throat disorders) presented the CRANIO registry: a registry focused on the outcome of treatment using standardised diagnosis specific outcome sets (starting with craniosynostosis and cleft lip/palate) including patient-reported outcome measures and outcomes on patients' quality of life. While substantial progress has been made in the development of a registry that will contribute substantially to better care and quality of life for European patients, data-sharing remains a real hurdle, both within the Netherlands, and cross-border.

All three presentations are recorded and can be found on: https://vsop.nl/actueel/satellietmeeting-vsop-tijdens-ecrd-conferentie/

DANISH SATELLITE MEETING

Approximately 40 participants joined the Danish Satellite Meeting, including patient advocates, clinicians and representatives from the Danish Health Authority and National Board of Social Services.

The Danish National Strategy for rare diseases was the main focus of the meeting, with most recommendations currently underway. The participants also discussed various topics related to accessing treatments: the need for centralised treatment for ultra-rare diseases, the importance of advanced therapy medicinal products, and the need to make it more attractive to include Danish patients in clinical trials. European Reference Networks were highlighted as a big success story, with further interest in joining other European initiatives such as Horizon Europe and the Rare Disease Research Partnership. Last but not least, the meeting stressed the need for sustainable funding solutions for the Helpline of Rare Diseases Denmark and the rare disease information point hosted by Lægebogen/sundhed.dk.

Key commitments included:

- Conclusion of a generic model for rare disease healthcare pathways in 2022
- External evaluation of the Danish National Strategy for rare diseases in the fourth and last quarter of the year 2022.
- Meetings in the advisory group for the Strategy before and after the evaluation.

SÄLLSYNTA DIAGNOSER
RARE DISEASES SWEDEN

SWEDISH SATELLITE MEETING

The Swedish Satellite Meeting took the opportunity of the upcoming elections to bring together policy makers from all eight political parties for a "Rare Debate". The need for more coordination of health care for people living with a rare disease in Sweden, both nationally and between Sweden's 21 regions, was one of the key outcomes of Rare Diseases Sweden's election debate.

The current situation in the rare area was described as "a symphony orchestra without a conductor, where no one holds the rare baton". The country's success in implementing policies that improve the situation of people with rare health conditions was evaluated as moderate. This was due to the fact that while there is indeed broad support for improvements, there is reluctance from the governing party to have a national strategy for rare health conditions. More coordination in primary care and more equal care throughout the country are also needed, as is the need to be more in line with patients' needs.

Rare Diseases Sweden shared figures from their latest membership survey, which showed that three out of ten respondents were denied care for simple conditions, due to their rare disease. Most politicians were appalled by the figures. Participants agreed that the problem is already well-defined, and has been for many years. What is needed is "to think outside the box on rare diseases", to quote Margareta Fransson from the Green Party.

The main problems to remedy were identified by Anders W Jonsson and included coordination - both locally and nationally - and access to orphan drugs. Different models, such as the Cancer Strategy model, were referenced to consider as a basis to move things forward. It was suggested that the authority had to bear the main responsibility, for example, the National Board of Health and Welfare, but that other authorities also needed a clear mandate to collaborate and become involved in the work.

MOBILISING THE RARE DISEASE MOVEMENT FOR UKRAINE: EURORDIS AND OUR ALLIES' RESPONSE TO THE WAR IN UKRAINE

What are the needs of people living with a rare disease in/fleeing Ukraine?

- Access to treatments;
- Access to care and medical consultations;
- Psychological support services;
- Basic needs such as food, accommodation, hygiene products and financial means.

On top of that, there is a need to shift the thinking from only addressing the urgent needs and covering gaps to also addressing mid-and long-term priorities, such as:

- Connecting Orphanet to the Ukrainian digital healthcare system and initiating the mapping of rare disease patients in Ukraine
- Creating a network of centres of expertise on rare diseases in Ukraine
- Expansion of the newborn screening programme
- Extension of state-guaranteed treatments to more rare diseases among them SMA, Duchenne and Tuberous sclerosis (including the promotion of registration of innovative medication in Ukraine)

Highlights of current and future solutions and strategies put in place to help people living with a rare disease in/fleeing Ukraine:

- Airbnb.org -EURORDIS partnership, which allows accessing and supports the most vulnerable populations with housing
- Razem z Ukrainą (Together with Ukraine) - a Eurordis-led confederation of 12 patient-focused non-profit organisations working together to meet the critical needs of a minimum of 500 Ukrainian families living with a rare disease who are currently in, coming to, and/or going through Poland.
- A Rare Diseases Virtual Hub as a dedicated focal point for any Ukrainian people affected by the war who needs specialised medical support for their rare and/or complex condition and to help this person and their family to better navigate the support systems available to them.

DR IRYNA MYKYCHAK
Deputy Minister of Health, Ukraine
National Alliance for Rare Diseases

> As a result of the war, 30% of patients with rare diseases have left Ukraine. We appreciate the support of Poland, the President of Poland and the people, to take care of Ukrainians. We are not refugees there, we are guests there, and we feel that. We appreciate the help of other EU countries helping our patients as well. We will never forget this.

HUMANITARIAN CRISIS IN UKRAINE

CALL TO ACTION:

For individuals:

- Donate
- Become an Airbnb.org host
- Support displaced Ukrainians

For decision-makers:

- Empower patient organisations and initiatives directly supporting ULWRD (Ukrainians living with a rare disease)
- Include ULWRD in your response by taking into account their complex needs (acceptable housing or transport; expensive medication)

For the industry:

- Support the healthcare sector of Ukraine to cover urgent gaps
- Promote more sustainable solutions for the accessibility of your medication

TOGETHER FOR RARE DISEASES

Multi-stakeholder initiative to unlock European Reference Network (ERN) collaboration with industry

PARTICIPATION

Attendees included representatives from ERNs, industry, patient groups and advocates.

MAIN TOPICS OF DISCUSSION, CHALLENGES AND OPPORTUNITIES

Sheela Upadhyaya, Chair of the Together4RD Steering Group, began the session with an outline of Together for Rare Diseases' (Together4RD) objective: to unlock collaboration between European Reference Networks (ERNs) and industry in areas that will address the 95% of unmet need in rare diseases. To date, collaboration between ERNs and the pharmaceutical sector has been limited and is further constrained by existing guidelines from the Board of Member States for ERNs.

The session opened with a keynote speech from Together4RD MEP Champion, Ondrej Knotek. Mr Knotek recalled the recent evaluation of the Cross-Border Healthcare Directive, which considered how to make ERNs more accessible. He stated how timely Together4RD's work to address unmet need for people living with rare diseases is.

A Panel discussion followed, where representatives of the Together4RD Steering Group (Professor Helene Dollfus, ERN-EYE; Toon Digneffe, Takeda; Ines Hernando, EURORDIS) called for action to address the barriers to collaboration. They explained the role they see for ERNs in rare disease research & innovation, and what the win-win outcomes are for ERNs and industry from closer partnership. The value that the patient community sees in Together4RD's work was also outlined. The panel discussion finished with a conversation around the future ambitions for Together4RD, and the collaboration pilot projects that will be used to demonstrate proof of concept to the European Commission and Board of Member States.

MAIN CONCLUSIONS

The need for Together4RD to unlock ERN and private actor collaborations was underlined by participants. Feedback from attendees was that the patient perspective must be reflected in decisions around the prioritisation and selection of pilot projects.

NEXT STEPS

- The Together4RD Policy Asks to unlock ERN and industry collaboration will be launched at an in-person event in the European Parliament in Brussels (10 November), which will be open to the wider community to attend.
- A White Paper summarising findings gathered by Together4RD on the barriers and opportunities for ERN and industry collaboration will be published at the end of the year.
- Pilot projects are anticipated to commence in 2023.

RARE DISEASES INTERNATIONAL

RARE DISEASES INTERNATIONAL (RDI)

Preparing the foundations of a Global Rare Disease Network

PARTICIPATION

Over 140 participants joined the session including patient advocates, clinicians, researchers, industry partners and policy leaders.

MAIN TOPICS OF DISCUSSION, CHALLENGES AND OPPORTUNITIES

Rare Diseases International (RDI) is collaborating with the World Health Organization (WHO) 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. The session explored how healthcare systems in the WHO European region could connect into a Global Network for Rare Diseases, building on and scaling up the European Reference Networks (ERN) system, exploiting the digitalisation of healthcare and harnessing the collective knowledge from the existing networks to unify an international expert community.

The main topics discussed the development of the ERNs and their international collaborations with the EMRO region. In particular, the session highlighted the partnership between ERN eUROGEN (www.eurogen-ern.eu) and CureforU (www.cureforu.com), a network initiative that supports virtual case reviews of infants with a rare urogenital disease in Pakistan, Afghanistan and Uzbekistan who need highly specialized surgery with experts from these countries connecting with experts from the ERNs.

Main challenges identified included securing funding to support the global network, e.g. seed funding for digital activities, support from regional and national authorities, and grants, donations and sponsorship. The session also highlighted the need to develop a sustainability strategy from the outset of the initiative.

MAIN CONCLUSIONS

The session reaffirmed the need for a Global Network for Rare Diseases. A network of networks in which clinical experts can offer their expertise anywhere in the world to help with the right diagnosis, treatment plan and share learnings from the ground and technology coming from different countries.

One insight gained was to leverage the tools, infrastructure and resources of existing mature networks to support the development of new networks and collaborations. For example, to explore how the ERNs can provide a learning and knowledge exchange space for other regions to learn from their experience in setting up new networks in the EU.

The session highlighted the need for RDI to support local and regional communities to be able to apply to join the pilot network, which is expected to be organised in the coming year(s), for example: supporting the piloting of new regional hubs in low-and-middle-income countries and regions.

NEXT STEPS

The next step is to explore, with the European Commission and the ERN Coordinators Group, the opportunities and activities that can be undertaken now to support the development of the pilot network following the UN Resolution on Addressing the Challenges of Persons Living with a Rare Disease and their Families. The Resolution encourages Member States to foster the creation of networks of experts and multidisciplinary specialized expert hubs for rare diseases, and strengthens international collaboration on research and data sharing.

RARE DISEASES (RD) PARTNERSHIP (HORIZON EUROPE)

Ambition, Vision and Mission

In a resource-limited context collaboration and synergy ought to be sought with various stakeholders, programmes, and initiatives to continue building a Rare Disease (RD) Ecosystem that leaves no one behind. The Rare Diseases Partnership concept paper details the missions and the first activity directions to (1) support a robust patient need-led research, (2) develop new therapies, diagnostic methods and pathways, (3) lead a digital transformational change connecting the dots between care, patient data and research, (4) ensure strong alignment of strategies in RD research across countries and regions and (5) structure a goal-oriented public-public and public-private collaborations targeted at interventions all along the Research & Innovation value chain.

This ambition set forth would require addressing the current RD research needs within, and beyond the RD Partnership through collaboration and synergies. To start identifying such collaborations, ECRD 2022 participants were invited to provide insights on three clusters: (1) topics to address within the RD Partnership, (2) topics to address through collaboration with industry, and (3) topics to address through synergies with other Partnerships / EU missions. This was held as an interactive exercise gathering +90 participants using a MIRO Board segmented into two scopes (a) fundamental and pre-clinical research investment to maximise the coverage of the 95% understudies RDs, and (b) Clinical research network, boosting the research capacity of the ERNs & other research consortia and hastening clinical trials development.

Many of the organisations, programmes, Horizon Europe Partnerships identified for possible collaboration in the RD partnership concept paper were pointed out by the participants for the scope on fundamental and pre-clinical research. Additional projects and platforms to consider for collaboration were provided. Newborn screening was provided as a topic of synergy between partnerships and EU Missions. The review of financial incentives (on an EU level) for the commercial sector to support the development and affordability of RD treatments was proposed to be tackled through collaboration with industry and in synergy with other partnerships and EU missions. Innovative therapies and/or Orphan Drugs development and investment were proposed as topics for industry collaboration leveraging on screening technologies, FAIR Data (from academia and industry) and AI, as well as pre-clinical research regulatory science and economic burden studies. Patients' and family's empowerment and involvement within the partnership as equal partners for research project design, application and review was highlighted, with an indication to exploit the patients' experience with healthcare (and within ERNs). The process of such topics was put in the intersection of the three clusters. Methodological support and Ethics advice for research were cited and both an overall process and workable policy that stimulate and enables the re-use of results were proposed. Drug repurposing was another of the main topics put forward to be performed within the RD partnership and in collaboration with industry, this is linked with personalised medicines collaboration, off-label use of drugs as captured by registries and the need to work more on legislation regulation.

Most of the topics proposed to boost research capacity of the ERNs & other research consortia and to accelerate clinical trials development were proposed to be implemented within the RD Partnership, whether in relation to the registry/databases exploitation, expansion of the network of European hospitals or setting their governance model. Collaboration with SMEs and industries to collect data through mobile devices in clinical studies was proposed, as well as to foster data use for diagnostic research. The Financial support for Health Care Providers to conduct clinical studies or maintain updated patients' registries was proposed as a topic of synergy with other partnerships and EU missions and with industry. At the intersection of the three clusters, the innovative clinical trials protocols and the implementation of Patients Centred Outcome Measures were put forward along with the logical collaboration with EMA. Reference was made to many of the topics proposed in the first scope (fundamental and clinical research) as there is an overlap and a continuum between the two scopes.

The full results of the MIRO board exercise are available through the following link: https://miro.com/app/board/uXjVOqCItEo=/?share_link_id=285336151886

These results will serve for the development of the Strategic Research and Innovation agenda of the RD Partnership that includes the aforementioned types of collaboration with the aim that the journey from knowledge to patient impact is expedited.

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]
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BACKGROUND

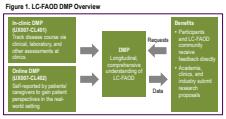
- Long-chain fatty acid oxidation disorders (LC-FAOD) comprise a group of rare, autosomal-recessive disorders of fatty acid metabolism in the mitochondria[1]
- These disorders may manifest in infants, but also commonly present in older children or adults following periods of increased metabolic demand, 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 (~19,300 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

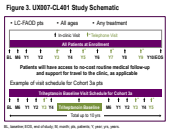
- Age-appropriate assessments (**Table 2**) will be performed in the DMP, depending on the patient's developmental age and physical ability at the time of the visit (**Figure 3**)
- Data for these assessments will be collected only if performed as part of standard of care for in-clinic visits

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 • Echocardiogram • Cardiac 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

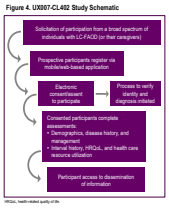


Online DMP (UX007-CL402)

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

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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

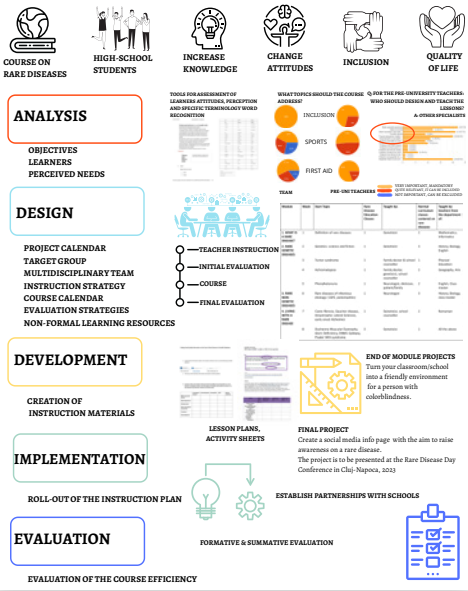
- The Norwegian welfare system is based on the local services.
- Impossible to maintain sufficient knowledge about rare diagnosis in every municipality
- TRS is one out of nine resource centres for rare diseases in Norway. The centres are established to:
  - Build knowledge about the diagnosis
  - Prepare 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.

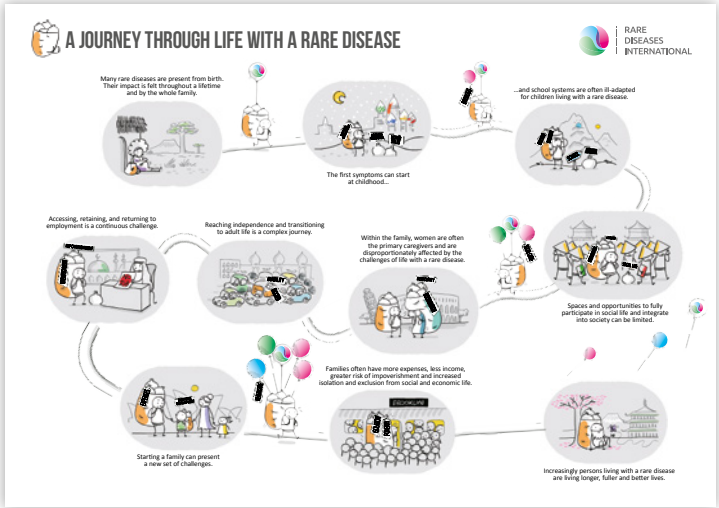
3RD PLACE

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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RARE DISEASES

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from diagnosis to cure

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rare diseases czech republic

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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
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Urogenital Diseases (ERN eUROGEN)

European Reference Network
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Coordinator
Assistance Publique - Hôpitaux de Paris, Hôpital Pitié-Salpétrière — France

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

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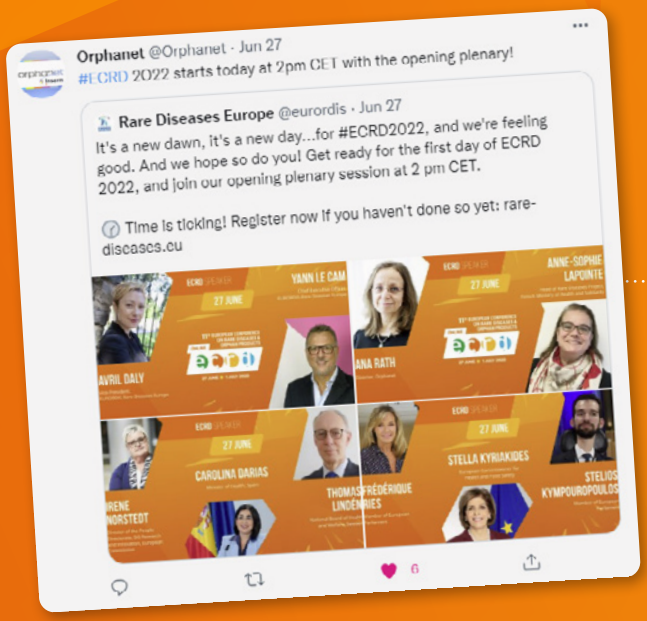
Orphanet @Orphanet · Jun 27
#ECRD 2022 starts today at 2pm CET with the opening plenary!
Rare Diseases Europe @eurordis · Jun 27
It's a new dawn, it's a new day...for #ECRD2022, and we're feeling good. And we hope so do you! Get ready for the first day of ECRD 2022, and join our opening plenary session at 2 pm CET.
Time is ticking! Register now if you haven't done so yet: rare-diseases.eu
YANN LE CAM
ANNE-SOPHIE LAPOINTE
AVRIL DALY
ANA RATH
CAROLINA DARIAS
STELLA KYRIAKIDES
IRENE NORSTEDT
THOMAS FRÉDÉRIQUE LINDEMANS
STELIOS KYMPOUROPOULOS


SMA-Europe @SMAEurope · 20t
We are excited to be a part of the #ECRD2022. Check out our posters: 125 on the Ukrainian SMA registry and 253 on SMA NBS Alliance. bit.ly/3ubirs2 @eurordis #RareDisease #SpinalMuscularAtrophy
11TH EUROPEAN CONFERENCE ON RARE DISEASES & ORPHAN PRODUCTS
ONLINE
27 JUNE 1 JULY 2022
MISSION POSSIBLE
PUTTING RARE DISEASE POLICY INTO ACTION


Medics4RareDiseases @M4RareDiseases · 27. jun.
M4RD are attending #ECRD2022 Good luck to everyone involved and thank you all for sharing your stories #raredisease @eurordis
PUTTING RARE DISEASE POLICY INTO ACTION


RDI @rarediseasesint · 21. jun.
WHAT IS A RARE DISEASE ⁉
Find out how the global community is working together to create an international description of rare diseases.
Don't miss the prize-winning poster!
#ECRD2022 rare-diseases.eu/register/
@eurordis @marywangCM @IrdIrc @Orphanet @black @AfmPresse
LEAVE NO-ONE BEHIND
UN RESOLUTION
What is a Rare Disease?
POSTER PREVIEW
Challenges from Rarity
Burden of Rare Disease
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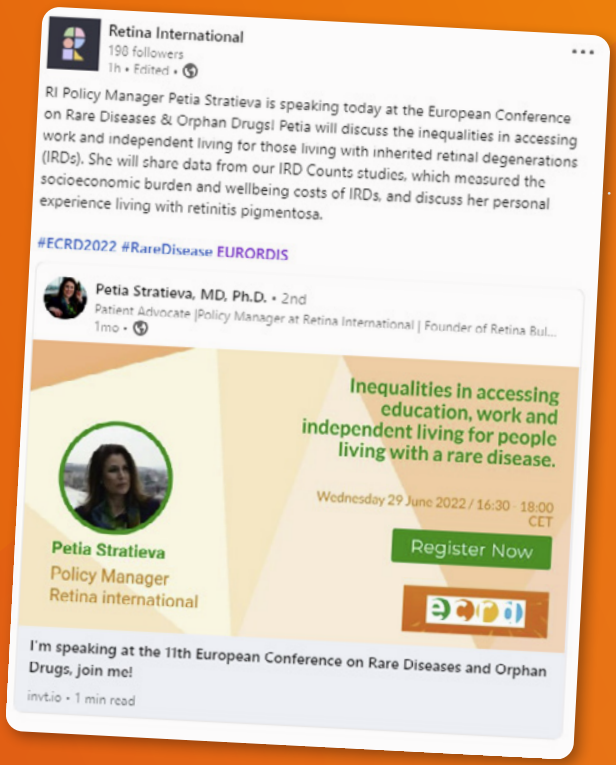
Retina International
190 followers
1h • Edited •
RI Policy Manager Petia Stratieva is speaking today at the European Conference on Rare Diseases & Orphan Drugs! Petia will discuss the inequalities in accessing work and independent living for those living with inherited retinal degenerations (IRDs). She will share data from our IRD Counts studies, which measured the socioeconomic burden and wellbeing costs of IRDs, and discuss her personal experience living with retinitis pigmentosa.
#ECRD2022 #RareDisease EURORDIS
Petia Stratieva, MD, Ph.D. • 2nd
Patient Advocate |Policy Manager at Retina International | Founder of Retina Bul...
1mo •
Inequalities in accessing education, work and independent living for people living with a rare disease.
Wednesday 29 June 2022 / 16:30 - 18:00 CET
Register Now
Petia Stratieva
Policy Manager
Retina international
I'm speaking at the 11th European Conference on Rare Diseases and Orphan Drugs, join me!
invt.io • 1 min read


Retina International
20 hrs •
RI Policy Manager Petia Stratieva is speaking today at the European Conference on Rare Diseases & Orphan Drugs!
Petia will discuss the inequalities in accessing work and independent living for those living with inherited retinal degenerations (IRDs). She will share data from our IRD Counts studies and discuss her personal experience living with retinitis pigmentosa.
#ECRD2022 #RareDisease EURORDIS - European Rare Diseases Organisation
14:00 – 16:00
Invisibility as a roadblock towards reducing inequalities
TRACK C/D
16:30 – 18:00
Inequalities in accessing health and social care
TRACK C
16:30 – 18:00
Inequalities in accessing and remaining in education & employment
TRACK D
EURORDIS - European Rare Diseases Organisation
21 hrs •
How can we improve the recognition, understanding and knowledge of rare diseases and their impact on the people who live with them?
Today, on Day 3 of #ECRD, ... See more
2
1 share


Veronica Lopez Gousset, MPH @vronnielolo · 20m
Come show my #poster some love! Top 10 common #needs of the #raredisease community around the world at #ECRD2022 DAY 3!
ONLINE
0:06 | 12 visninger
27 JUNE 1 JULY 2022
2


EuRR-Bone @EuRRBone · 27. jun.
Are you attending #ECRD2022? Find our ePoster presented by @InesA_Beyond and more at the meeting platform. The European Registries for Rare Bone and Mineral Conditions (EuRR-Bone) the Use of a Core Registry to Collect Patient and Clinician Reported Outcomes
The European Registries for Rare Bone and Mineral Conditions (EuRR-Bone): the Use of a Core Registry to Collect Patient and Clinician Reported Outcomes
EuRR-Bone
European Registries for Rare Bone and Mineral Conditions
Abstract
Poster
Newsletter
4
10


ZoiAn @ZoiAn2 · 28. jun.
Day 2 of #ECRD2022
Invisibility as a roadblock towards reducing inequalities.
Sandra just shared results of a Rare Barometer survey, I cannot wait for it to be published, to share it with you.
#RareDisease #diagnosticoddyssey #patientjourney #vasculitis
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ONLINE
27 JUNE 1 JULY 2022
5


International Society of Nephrology (ISN) @ISNkidneycare · Jun 27
On the opening day of the #ECRD2022, and as outlined during #ISNFrontiers, the ISN reiterates the importance of diagnosis and innovative treatments to improve the lives of ~2 million Europeans suffering from one of 300 rare #kidneydiseases.
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PUTTING RARE DISEASE POLICY INTO ACTION
Rare Diseases Europe
3
8


EUCOPE @EUCOPE · 20h
"We also don't want to fall behind the US in terms of OMP incentives." EUCOPE's Alexander Natz highlights the differences in order for the EU to maintain competitiveness with the US. #ECRD2022
Making Europe attractive for therapies development for PLWRD - addressing the valley of death
"In the EU, we have a very different situation where after EMA approval, there are 27 different negotiations to go through and additional time lags to achieve reimbursement in comparison to our US counterpart."
Alexander Natz
Secretary-General
EUCOPE
#ECRD2022
Rare Diseases Europe and 2 others
1


The TAPS Support Foundation · Twin Anemia Polycythemia Sequence
27 June at 14:51
While our research team are working hard representing twins at #FMF2022, we're representing our #raredisease side at #ECRD2022. Looking forward to learning and listening in some incredible sessions. We're also poster presenters! Check out our video poster here: https://bit.ly/3A7G5lb learn about our amazing collaboration with @fetallumc. #tapssupport #tapsisreal
#ECRD 2022
ONLINE
27 JUNE 1 JULY 2022


Therapeutic Advances in Rare Disease @TARareDisease · 19h
Attending #ECRD2022?
Make sure you tune in to the next session 'How to make the best use of ERNs & improve the efficiency of the whole system'
Chaired by our Associate Editor @maurizioscarpa
Today! 16:00 – 17:30 CET
@eurordis @Metab_ERN
#RareDisease #ECRD
27 JUNE 1 JULY 2022
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