HYBRID ecrd

ACTION WITHIN REACH

PIONEERING SOLUTIONS FOR RARE DISEASES

HYBRID 15 & 16 MAY 2024

12th European Conference on Rare Diseases and Orphan Products

EXECUTIVE **SUMMARY**

Under the auspices of

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ORGANISED BY

EURORDIS RARE DISEASES EUROPE

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#ECRD2024

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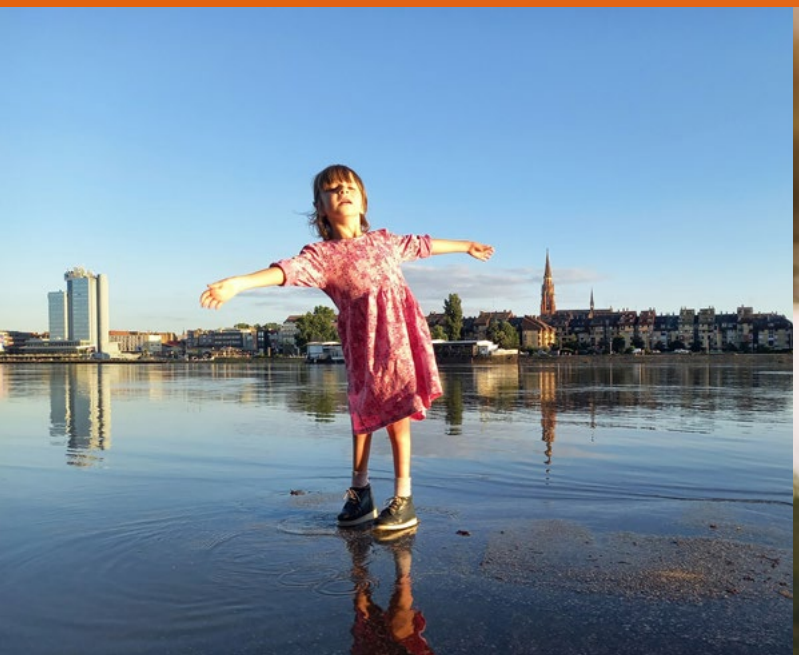

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KEY FACTS AND FIGURES

49 countries represented

700+ participants

14+ **Ministers, MEPs, European commission** representatives

4 **Pre-Conference Webinars**

50+ **experts** as session chairs, speakers & panelists

14 **plenary, Parallel and Small Group** sessions

87 partners

130+ ePosters displayed (with 30+ displayed in-person)

5.9 K+ **people reached** on Instagram

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 takes place, forming the groundwork to shape future rare disease policies.

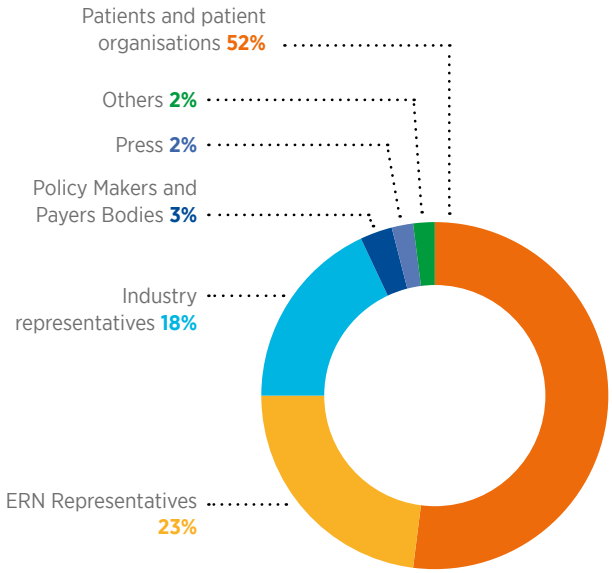
The 12th edition of the European Conference on Rare Diseases and Orphan Products (ECRD) was fully hybrid for the first time. This new format ensured the conference remained accessible for all, whilst also providing the opportunity for stakeholders to come together in Brussels ahead of the 2024 European Parliamentary elections. The event had a combined audience of over 700 participants from across 49 countries.

Throughout the two-day programme, participants were able to take a deep-dive into discussions on how EU institutions and country leaders can keep rare diseases as a key focus within healthcare policy. A number of pre-conference Thought Leader Sessions, alongside a library of resources including podcasts, videos and research papers, were organised in advance of the conference to bolster the learning experience for registered and prospective attendees.

With the help of our custom-built platform and professional conference facilitators, the online and onsite participants were brought together to engage in a range of keynote sessions, learning from the insights and testimonies of over 50 expert speakers and chairs. All participants played an active role through joining networking sessions, contributing to small group discussions and exchanging knowledge through a hybrid display of over 130 posters.

This year's conference culminated in the co-signing of the EURORDIS Open Letter; an official summary of the policy asks and key-takeaways from ECRD 2024, to be presented to the European Commission.

STAKEHOLDER BREAKDOWN



SETTING THE STAGE FOR ECRD 2024

This year's **ECRD** coincided strategically with the upcoming European Parliament election, providing an opportunity to leverage this timing to push for a comprehensive policy framework, addressing the needs of the rare disease community. The upcoming elections and the subsequent appointment of the new leadership of the European Commission have been critical, determining the future direction of EU policies, including those affecting rare disease research, funding, and healthcare access.

Building on recent milestones, such as the United Nations' Resolution on ''Addressing the Challenges of Persons Living with a Rare Disease and their Families'' and with the robust support from European policymakers, the conference aimed to transform these acknowledgements into actionable strategies to address persistent inequalities and unmet needs across the continent.

ECRD 2024 discussions were inspired by the eight key areas detailed in EURORDIS' '**Championing the Rare**' manifesto (**#ActRare2024**), including the development of plans for rare diseases at national and European levels, innovative research funding strategies, enhanced mental health support, and reduced diagnostic delays.

Another significant feature of this year's conference was its hybrid format, combining in-person sessions in Brussels with virtual participation. This facilitated broader engagement and ensured that diverse perspectives and experiences were represented.

The main outcome of the conference was the launch of an Open Letter to future EU policymakers. This letter advocates for a European Action Plan for Rare Diseases and calls for coordinated policy actions across EU Member States to ensure equitable access to diagnosis, treatment, and care. It represents a collective call to action, urging policymakers to commit to a unified strategy that addresses the multifaceted challenges faced by the rare disease community.

LEVERAGING POLITICAL MOMENTUM FOR 2030 GOALS

Despite international recognition of the challenges faced by the rare disease community and commitments from policymakers, progress over the past decade has been insufficient. The gap between policy declarations and real-world outcomes is widening, signalling the need for immediate and concrete actions.

ECRD 2024 highlighted the urgent need for accelerated efforts to meet the recommendations of **Rare 2030**. Developed with the UN's ambitious 2030 Sustainable Development Goals as the backdrop, these recommendations emerged from a collective effort of the rare disease community at large and continue to be the compass for EU policy action for people living with rare diseases and their families. With less than six years remaining, the current slow pace of progress threatens to leave the community's needs unmet.

The conference identified bottlenecks hindering progress and noted that the rate of advancement in research funding, policy implementation, and healthcare delivery has not kept pace with the community's escalating needs. It therefore proposed concrete actions 'within reach' that could overcome these barriers and address the most pressing needs of the rare disease population.

ECRD 2024 served as a wake-up call to all countries regarding their commitments towards 2030, urging immediate and coordinated efforts to ensure that people with rare diseases receive timely, equitable, and comprehensive care.

It also provided the new cohort of policymakers with a roadmap of incisive actions that have the potential to improve the lives of people with rare diseases.

Harnessing political momentum is essential for driving substantial and lasting enhancements in rare disease policies and practices.

KEYNOTE SPEECHES

FRANK VANDENBROUCKE
Minister of Health and Social Affairs, Belgium

To improve the situation of those 30 million Europeans, collaboration beyond borders is crucial. A European Action Plan on Rare Diseases can contribute significantly to improving that collaboration. We should aim for a situation where people with a rare disease have the same chances independently of where they live.

AVRIL DALY
President
EURORDIS- Rare Diseases Europe

Being together really provides the creativity that can shape the direction of our policies, not only in the current ecosystem we are living in now but also for future policies.

VIRGINIE BROS-FACER
Chief Executive Officer
EURORDIS-Rare Diseases Europe

Across 12 editions, we have experienced how ECRD has become a key driver in shaping policies. We believe that today, again, we can collectively spell out solutions to address the unique needs of people living with rare diseases and their families and shape the policies of tomorrow.

LUCY MCKAY
CEO
Medics4RareDiseases

We need to make sure that the world is psychologically a safe place for people living with rare conditions. [...] If you think that your knowledge and experience would be valuable, which it will be, make yourself heard.

STEFAN JORIS
Chair
Association Muco

If you want to address the needs of very small populations with specific, high unmet needs, you need to collaborate across borders. [...] [Country-level] is not enough if we want equitable access to care and medication across the different Member States. There's no other way.

ANA RATH
Director
Orphanet

This is the starting point of the next political cycle for rare diseases in Europe. It took nearly 24 years to set the solid ground on which we will build the future for rare diseases. We must ensure that rare diseases remain a priority for the next European Commission.

KEYNOTE SPEECHES

EVA SCHOETERS
Director
RaDiOrg

We need to advocate for mental health support to be an integral part of rare disease care, making it a key part of standard care. We call on the European Union to support Member States in developing effective programmes to make this a reality.

VALENTINA BOTTARELLI
Public Affairs Director & Head of European Advocacy, EURORDIS

We need an overarching framework in the form of an action plan to unite ongoing and future efforts in a comprehensive way. This plan must bridge diverse policy areas, streamline efforts, and set clear, measurable objectives for our collective direction. This is a call from the broader health community.

SOFIE SKOUBO
Paralympic Athlete and PhD Student

Education is a fundamental pillar of society and a lifeline for those whose physical capabilities limit their career options. Society must provide the necessary support to ensure children with chronic illnesses can access education.

STELLA KYRIAKIDES
European Commissioner for Health and Food Safety

The EU shares EURORDIS' overarching goals for rare diseases: to improve patient access to diagnosis, treatment, and care. [...] Nobody can be left behind. People suffering from rare diseases must have access to the best, the most innovative treatments, no matter where they live, and no matter their disease.

ALAIN COHEUR
President, Section for the Single Market, Production and Consumption, European Economic and Social Committee

Over the past two years, [we have worked] on building trust and sharing a vision to establish a comprehensive European action plan on rare diseases, to ensure that all rare disease patients in the EU have equal access to diagnosis, treatment, and holistic integrated care. Now we have a roadmap – let's start.

JAKUB DVOŘÁČEK
Deputy Minister of Health, Czech Republic

We need a joint plan for rare diseases. We must seize the opportunity with the new European Parliament and the new Commission to build on this momentum. Europe needs to focus more on health.

THE OPEN LETTER

ECRD 2024 concluded with a collective call to action, stressing the urgency for European leaders to prioritise health in future policies and deliver policy solutions for people living with rare diseases.

An Open Letter to the European Commission was crafted, pushing for a comprehensive European Action Plan for Rare Diseases and the implementation of immediate actionable measures in upcoming work programmes to address the critical needs of the rare disease community.

Throughout the two-day conference, actionable policy recommendations were formulated that can be incorporated into a unified framework under a European Action Plan.

These recommendations focus on enhancing EU and national plans for rare diseases, improving diagnostic processes, accelerating innovative research, and ensuring equitable access to treatments and highly specialised care.

KEY REQUESTS IN THE OPEN LETTER INCLUDE:

1. Maintain health at the forefront of future policies and programmes.
2. Develop a comprehensive European Action Plan for Rare Diseases that bridges diverse policy areas and streamlines existing efforts with clear, measurable objectives.
3. Immediately address the most pressing needs of the rare disease population by integrating the actions 'within reach' that we have collectively identified within the work programmes of the next years.

At ECRD 2024, the rare disease community converges, ready to spark a new wave of advocacy and action.

Voices rise in unison, calling for a European action plan that outlines the path forward in areas where progress is most urgently needed.

KEY POLICY ASKS

EUROPEAN AND NATIONAL POLICY FRAMEWORKS FOR RARE DISEASES

Establish a European Action Plan for Rare Diseases, an overarching framework with clear, measurable objectives. Within this framework, support collaborative actions across EU Member States and other European countries, notably to:

- Identify common goals and develop an integrated system of indicators that enables to inform, shape and constantly improve decision-making, policies and systems at the EU and national level;
- Create an EU-wide multistakeholder body to steer and oversee the above collaborative actions, thereby streamlining efforts and ensuring consistency in care and support for people with rare diseases throughout Europe.

EARLIER, FASTER AND MORE ACCURATE DIAGNOSIS

Promote EU-level initiatives that enhance cross-country cooperation on newborn screening. This will bring newborn screening national programmes across Europe closer together and make them equitably accessible. Align on best available standards, methodologies and scientific advancements, or issue recommendations to expand the scope of rare diseases in those programmes, among others to increase chances to access further treatments.

INNOVATIVE AND NEEDS-LED RESEARCH

Create a European development platform and explore novel models to speed up clinical research and development on very rare and underserved diseases. This must be coupled with ambitious investment funding strategies, both from the private and public sectors.

TIMELY ACCESS TO AFFORDABLE AND INNOVATIVE TREATMENTS

Unravel the paradox of patient access to rare disease therapies in Europe, notably by:

- Exploring the feasibility of novel payment mechanisms, from subscription models to a common EU access pathway and cross-country negotiations;
- Creating a common approach to funding treatments for very small populations based on a solidarity mechanism to provide access across borders.
- Enhancing collective action to ensure timely access to highly specialised healthcare:
- Creating a European system to centralise the commissioning and delivery in a few centres of highly specialised healthcare services for very small patient populations and certain highly specialised complex procedures.

INTEGRATED, PERSON-CENTRED AND LIFELONG HOLISTIC CARE

Support EU Member States to implement the United Nations General Assembly Resolution's (A/RES/76/132) call to develop effective programmes and national strategies to promote mental health and psychosocial support for people living with a rare condition.

Integrate psychological support as an essential component of holistic, patient-focused care, by enhancing existing medical care to be 'psychologically informed' for people living with a rare condition.

Recognise and support patient organisations to provide community and peer support, and access to trusted information, as the foundation of psychosocial care, enabling earlier detection and access to preventative support.

PARALLEL SESSIONS

REVOLUTIONISING FUNDING STRATEGIES FOR BREAKTHROUGH THERAPIES IN RARER DISEASES

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

THE PATH FORWARD FOR EQUITABLE DIAGNOSIS

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

INNOVATING THERAPIES, UNEQUAL ACCESS BRIDGING THE GAP FOR RARE DISEASE TREATMENTS

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

REVOLUTIONISING FUNDING STRATEGIES FOR BREAKTHROUGH THERAPIES IN RARER DISEASES

This session highlighted the challenges involved with funding treatments for rarer diseases. As explained during the session itself, there is no clear definition for rarer, ultra-rare, very rare and nano-rare diseases. **Around 85%** of rare diseases have a prevalence of **less than 1 in 1 000 000** whereas around 80% of rare disease patients are affected by the 150 most prevalent diseases **(1 to 5 per 10 000)**. Very rare diseases are diseases with very few affected patients and families, with limited knowledge and a limited evidence base, not benefiting from specific incentives and facing additional regulatory challenges due to their extreme rarity. When discussing timelines and challenges relating to research in very rare diseases, quality development must be data-driven and adhere strictly to timelines. Also, early discussions with regulatory authorities are crucial. The rarer the condition, the more complicated it gets. Very rare diseases are commercially risky to invest in for drug developers because the average and current drug development funding process is not compatible with nano-rare/very rare diseases.

When approaching funding strategies for very rare diseases, not only the funding level and/or more funding should be considered but also how to fund new therapy developments differently. Different drug development processes have emerged and have proven to be effective for n-of-1 or n-of-few. Initiatives deviating from **“1 mutation, 1 medicine”** are also getting increasing attention. To develop translational systems, addressing groups of rare diseases is also an approach that is gaining momentum. It is becoming obvious that the traditional drug development process is not going to work in very rare diseases and that academic drug development might be an opportunity to pursue.

Corporate interest and success in rare disease therapeutics lags behind patient needs and demand. A recent survey conducted by the **IRDiRC Chrysalis task force** identified key financial and non-financial factors that make rare disease research and development attractive to companies. This survey highlighted attractive features and potential barriers for company involvement in rare disease, mostly within the context of “rarer” diseases.

Some of those opportunities include gaining a good understanding of the genetic cause of the disease and biological mechanisms leading to the pathology; the possibility to address a high unmet need; a collective expertise applied to a shared purpose; the quantification of disease burden and unmet need, as well as determination of risk–benefit calculus by patient communities.

Challenges included the small numbers of patients, the lack of or incomplete understanding of clinical progression, the lack of quantitative understanding of disease burden, the lack of reliable models of the disease, the lack of potential biomarkers for early clinical studies, the lack of global concordance on regulatory pathways, the lack of predictability of reimbursement negotiations as well as some misalignment between industry and academia or research institutions for effective partnerships.

Nevertheless, some funding opportunities are available, such as those from the **European Investment Bank** that supports mid- to late-stage development. This funding body supports policy-focused investments in critical sectors and technologies in Europe, complementing both public and private funding sources and generating a signalling effect and contributing to crowd-in investors. Other models need to be further investigated and developed, such as collaborations and partnerships, including de-risking science and platforms' approaches. Sharing data and information and unlocking the first steps of the science to move forward require further efforts and common initiatives. Bespoke therapy (treating several diseases with the same technology) is one positive example of how the rare disease community can work together.

When approaching therapy developments via platforms, transversal and global is thinking key. A deviation from traditional models is needed, together with the respective funding, to pursue those objectives which require funding from private and public sources.

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

The **"No Health Without Mental Health! Let's Co-create a Mentally Healthy Toolkit"** session was an official event of European Mental Health Week that ran from 13-19 May. The theme the 2024 campaign was "Better Together: Co-creating the Future of Mental Health".Our session launched a co-creation process to develop a new Mentally Healthy Toolkit for the rare disease community. We completed two exercises to identify the common needs faced by our community and then explored the existing approaches and tools, learning how they have worked for our community, and testing if they are applicable to form the basis for a new toolkit.

"Diagnosis and living with a rare disease have a profound impact on an individual and their whole family. The impact and supportive and protective benefit associated with a rare disease patient group existing should not be underestimated! By existing you provide a sense of belonging." Kym Winter, Rareminds.

The main conclusions that came through the session recognised the important role patient organisations play and the support they provide to people living with rare diseases, to access community and peer support, and access to trustworthy information. The session called for the **European Commission & Member States** to recognise (and support) patient groups' role in being a foundation of psychosocial care, enabling earlier detection and access to preventative support.

"Reducing isolation by meeting people who know your story is a huge milestone for people living with rare diseases. The first time you attended a conference where everyone knew about your rare condition is a game-charger in the rare disease journey. This created a feeling that everyone else was in my world and I was no longer alone" Eva Schoeters, RaDiOrg.

The four common needs highlighted from the session were:

1. Living with uncertainty and coping with change, grief and loss
2. Reducing isolation and connecting with others
3. Confidence to speak up and break down stigma
4. Self-care strategies

The toolkit will be developed specifically to address the common unmet mental health needs experienced by the rare disease community.

"Everyone who is touched by a rare or undiagnosed condition needs to be connected to a rare disease advocacy group. Once they get access to this beautiful community, they will never be alone. This sense of belonging is a protection factor for wellbeing and the key to breaking social isolation," Lucy McKay, Medics4RD.

Through engaging with the active participants, we started to build a picture of the toolkit, which needs to be disease agonistic to address commonalities experienced by the community, as well as be flexible and adaptive to condition-specific needs, and able to address the diverse needs of children, young people and adults living with a rare disease and their wider family.

"Reducing isolation starts with the announcement of a diagnosis, and indeed before the diagnosis. We need to reinforce the family's resources and provide strategies to support the whole family." Dorica Dan, Romanian National Alliance for Rare Diseases.

Based on existing tools and approaches that have been seen to work in different communities, we have identified 5 key domains that we will further elaborate on as a blueprint for the toolkit, including:

- 1) Self-care and support for where there are no existing patient groups.
- 2) Therapeutic community support including the role and value of disease-specific patient groups, access to trusted information, family weekends, etc.
- 3) Peer-to-peer support on an individual level including mentoring, individually tailored information and support etc.
- 4) Psychosocial care advocacy tools to secure access where there is a service gap.
- 5) Health and social care professional tools and approaches.

Building on this foundation of psychosocial support, we highlighted the need for the **European Commission and Member States** to support the development of effective programmes and national strategies to promote mental health and psychosocial support for people living with a rare condition. We see the implementation of psychosocial care for people living with a rare disease as an 'action-within reach' as this can be done by enhancing existing medical care to be 'psychologically informed'. This would fulfil the commitment in the United Nations General Assembly Resolution on tackling the challenges of people living with rare diseases and their families. Finally, the session also concluded that there is a clear and pressing need to make mental health visible in rare disease national plans & strategies. When national rare disease plans are updated, it is recommended that the plans should recognise the impact of rare conditions on mental health and include actions to address these needs.

THE PATH FORWARD FOR EQUITABLE DIAGNOSIS

As this session outlined, receiving a timely diagnosis is one of the most serious challenges faced by people living with rare diseases. The average total diagnosis time is still close to 5 years as evidenced by a **2022 EURORDIS-Rare Barometer survey**. Diagnosis is a pivotal point in the rare disease journey. While the term of a 'diagnostic odyssey' is commonly used, with **more than 7 specialists** consulted as an average to reach a correct diagnosis, it should not be overlooked that some people do not get to the end of the odyssey. Some of the many challenges along the way are getting lost in the healthcare system, the possibility of misdiagnosis, or remaining undiagnosed. Expanding newborn screening has emerged as one of the solutions to help shorten time to diagnosis, gaining strong support from people living with rare diseases as demonstrated by a **2023 Rare Barometer survey**. 90% of the respondents to this survey thought that any rare disease should be screened at birth if:

1. It allows a quicker diagnosis, to the benefit of the individual person and their family members;
2. It allows people living with rare diseases to have their disabilities better recognised, more adequate social support and independent living;
3. The disease can be followed-up and harm can be avoided through prevention practices.

As exemplified by Italy, early detection of rare diseases can dramatically change the course and quality of life.

Rare diseases, characterized by heterogeneity and geographical dispersion, are considered an emerging global public health priority that needs to be addressed by integrated healthcare systems. Genetic diseases are a top contributor to infant mortality. Several initiatives on newborn genomic screening have been launched globally to directly tackle this challenge.

The guiding principle for all those programmes is early detection and intervention which leads to better health outcomes, signalling a clear shift to preventive medicine.

Newborn genomic sequencing is considered as both a screening test falling in the domain of public health and a diagnostic test falling in the domain of precision medicine. Some disorders can be treated with new gene therapies prior to irreversible damage, but only if detected early. Newborn genomic sequencing could also bring more equity in the healthcare domain as it would concern all babies born, with no discrimination. Three key components are needed to implement newborn genomic sequencing: **feasibility, clinical utility, and societal acceptance**. All these need to be in place before a public health measure is put in place. In contrast to sporadic cancers, **up to 10%** of all cancers occur because of inherited genetic defects and this could be similar in all conditions.

With that context in mind, early genome sequencing in routine clinical care offers a quick route to diagnosis, particularly in combination with an interdisciplinary clinical workup. If this is embedded with genetic counselling prior and

after, it is well tolerated by patients. Genome sequencing fosters preventive care, family planning, testing of relatives, personalised management, and ending an odyssey. Genome data can also offer the opportunity to a further chance to be re-evaluated and to potentially benefit from a further diagnosis. Novel models like the **SWAN (Syndrome without a name) Clinic** also contribute to delivering a further chance for diagnosis to those with an unsolved disease after state-of-the-art investigations.

With a whole family approach possible, diagnosis could be made in patients without a diagnosis via re-analysis of genetic data and re-identifying of the phenotype. In those patients who have completed investigation and been discharged, the diagnostic rate rises to **35%**.

With patient-reported experience measures **(PREMs**), patient-reported outcome measures **(PROMS)** and patient interviews, improvements have been demonstrated in patient experiences particularly in care co-ordination. Patients appreciate the multi-disciplinary and holistic nature of the **SWAN clinic**, with a key role being held by the Clinical Nurse Specialist (CNS) Navigators.

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

The session discussed the opportunity to establish a European model to plan and commission certain highly specialised treatments. The following themes were covered through the discussion:

Inequities in access to cross-border highly specialised healthcare persist.

The EU cross-border healthcare system that's in place today does not ensure that everyone with the same condition and needs can access cross-border healthcare under the same conditions. To illustrate these inequities in access, 4 cases of people with anorectal malformations who had undergone surgery at Radboud University Hospital in The Netherlands were presented. In the best scenario, patients use the route of the Social Security Regulation (S2 form); their insurance fund or their national health system reimburses the treatment expenses. However, patients who do not get the authorisation for cross-border healthcare need to pay for the surgery out-of-pocket.

Inequities may be minimised or entirely avoided when a country has a well-functioning system for centralised commissioning.

The case of NHS England and its contract with UZ Leuven to perform open fetal surgery for spina bifida was presented to illustrate the situation. Patients have access, under the same circumstances, to the same level of care, either in England or in Belgium, depending on where they live. Importantly, the pathway has time-bound steps, there is no paperwork and patients get automatically referred.

Different arrangements and EU action is needed depending on the needs of patients, the prevalence or incidence of the condition, the size of countries and the number of Expert Centres who may perform the procedure with the necessary assurances.

In large and medium-size EU countries, for the most prevalent rare conditions and surgical procedures, at a country level some form of centralisation would be the first step. The EU could help by establishing standards and criteria for the Expert Centres that should be observed by all countries in their centralisation strategies. The criteria developed by the ERNs could be a starting point but should probably be reviewed.

For very rare diseases, rare surgical procedures with a very low EU-wide caseload or treatments delivered in a few centres, where it is very difficult to organise services at a national level, the EU should have a well-functioning system for cross-border healthcare, that would allow countries to plan services together, refer patients with the same condition to a few centres and cut the red tape.

First steps towards a European system for highly specialised healthcare.

First, countries need to define what highly specialised procedures would benefit from greater cooperation at EU level. EU countries could then define together service specifications, set expectations for what healthcare providers (HCPs) need to deliver, as well as the criteria that healthcare providers would need to meet. HCPs could be selected by an evaluation panel composed by

commissioners, patient representatives and clinicians. Follow-up would also need to be organised, either locally, in close coordination with the Expert Centre, or by having the patient travelling for follow-up, or a combination of both. With such a system, all patients living in the EU with a given condition and eligible for treatment would get referred automatically to the same centres.

The role of ERNs as treatment eligibility panels.

Regarding the role of the ERNs as "treatment eligibility panels" in such an EU system for highly specialised healthcare, the speakers highlighted the role of European expert panels in making joint decisions on complex cases.

Establishing criteria.

For rare and complex surgical procedures, the criteria to select these centres should always include caseload. Other quality criteria could refer to training and innovation capacities, research, sustainability of the service, patient outreach, etc., as well as hospital-related factors.

Partnership to progress on next steps.

Speakers agreed on the need to work together to define concrete steps to improve the current situation and facilitate patient access to highly specialised cross-border healthcare, specifically for very rare and complex surgeries or treatments that are delivered in a few centres in Europe. The EURORDIS working paper on a European Health Union for highly specialised healthcare was circulated after the session for comments and feedback.

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

As this session highlighted, the European Union is currently facing major challenges in ensuring equitable access to rare disease treatments. Although treatments for rare diseases are already limited, the few that do exist face significant disparities in access across EU Member States. In some countries, access to these treatments is faster, while in others there are significant delays. These disparities underline the urgent need for a coherent European solution that guarantees equal access for all citizens, regardless of their geographical location. As highlighted in Enrico Letta's report on the future of the EU's single market (and mentioned during the session), there is an urgent need for EU policies that bridge these gaps and ensure that no one is left behind.

The principle learning objectives for this ECRD 2024 session on **Innovative Therapies, Unequal Access: Bridging the Gap for Rare Disease Treatments** were as follows:

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

The critical question addressed during the session was how to implement better policies for better approaches within the EU. The consensus among speakers was clear: cooperation is key. Collaborative efforts at both a national and an EU level are essential to address the current access challenges. Key strategies discussed included increased transparency as well as joint EU negotiations as a promising strategy, particularly for ultra-rare diseases where collective negotiations can have a significant impact due to the small number of patients. The commitment of all EU and national stakeholders was also highlighted as crucial to make a tangible difference in improving access to rare disease treatments and ensuring that no patient is left behind.

The session also identified additional immediate steps that are needed to eradicate access issues. These include specialised administration, staff training, improved infrastructures and better access to and availability of diagnostic tests. The crucial role in this of the **European Reference Networks (ERNs)** was also discussed.

The session finally emphasised the importance of patient involvement from the outset to highlight unmet medical needs. Ensuring that patients are involved and that all stakeholders work together towards common goals will be essential to address inequalities in access to rare disease treatments across the European Union. Encouraging initiatives are already underway, but only through unwavering commitment can we ensure such a success.

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

The aim of this session was to bring a renewed focus on rare disease national plans (RDNP) by showcasing good practices developed in different countries that have yielded positive results and could be either replicated or tailored to other healthcare systems.

Almost all EU Member States - and many other countries besides - complied with the initial 2009 European-level request to adopt a national plan (NP) or national strategy for rare diseases; however, many issues remain, in terms of the status, detail, governance, funding, content and monitoring of national plans. The Rare 2030 Foresight study issued numerous recommendations here, one of which was to identify and share good practices: this ECRD took the first steps in this direction, by sharing several good practices developed through the national plans of Luxembourg Italy and France, concerning governance, monitoring and national healthcare networks, respectively.

These countries were represented by a duo of speakers from a national competent authority and a National Alliance of Rare Diseases, demonstrating an essential good practice, namely close collaboration between the rare disease patient community and health authorities.

Luxembourg has established a RDNP built upon patient-centric governance.

The RDNP Steering Committee and all Working Groups include representatives from ALAN - the National Alliance - other patient organisations and individual patients with a voice equal to other stakeholders.

Italy has a bottom-up, comprehensive RDNP monitoring programme.

The National Alliance, UNIAMO, initiated MONITORARE in 2015. Starting with a couple of basic indicators, this programme has expanded to include more indicators with a view to monitor and assess RDNP actions in more depth. MONITORARE collects data from all the actors involved in the Italian RDNPs.

France has structured RD healthcare networks involving patient representatives.

Over the past twenty years, within three successive RDNPs, France has comprehensively mapped rare disease medical expertise into over 600 highly specialised Centres of Expertise and 1,700 'Support Expertise Centres', grouped into 23 national healthcare networks.

The session then embarked on panel and full-group discussions.

The sharing of knowledge is crucial in rare disease. Countries should strive to learn from each other and tailor good practices to their national systems. For instance, patient-centred governance should be easily replicable in all RDNP and the MONITORARE programme can also be easily replicated (patients' involvement is important to select indicators that are the most relevant to them).

Although some countries have a dedicated national plan budget, many do not, and the absence of specific funds should not necessarily be seen as a barrier to developing plans or actions: funding can be drawn from budget lines in other existing health plans and services.

Participants agree that there has been a lack of focus at the European level on the topic of national plans, which has resulted in a lack of focus at the national level, and in some countries, lower incentives to maintain a dynamic, action-driven NP. It would be easier to develop robust RDNPs within a joint European framework that could help steer the process across countries towards shared common goals and facilitate the sharing of good national practices as well as the integration of European rare disease policies. It would be beneficial for European countries to develop an updated set of common monitoring indicators, which should be patient-centred, centred on the Sustainable Development Goals, and include ethics, autonomy and equity.

Dr Jakub Dvořáček, Deputy Minister for Health of the Czech Republic, emphasised that a European Action Plan for Rare Diseases should include the golden standards to be reached in all areas covered by a rare disease plan, stipulating the best way forward for Europe.

Conclusion

The recommendations set out in the Rare 2030 Foresight Study can serve as a good base for action here, at national and European level, and should be implemented as soon as possible. Key amongst these is the provision of an official multistakeholder European forum to support learning across countries and discuss rare disease policies broadly, beyond ongoing thematic EU projects. The rare disease community truly hopes that the new EU Parliament and Commission will place rare diseases as a public health priority and vote for the adoption of a European Action Plan for Rare Diseases called for by the community.

SATELLITE MEETING
Organised by RONARD

On 13th May 2024, the Romanian National Alliance for Rare Diseases organised a Satellite Meeting to complement the focuses of this year's ECRD. The meeting took place under the theme of the Connection of European activities for Rare Diseases with the National Plan for Rare Diseases and integration into the national health care system.

The meeting welcomed representatives from across the rare disease community, including patients, experts, alliances and organisations, and representatives from the Romanian national authorities.

The goal of the ECRD satellite meeting was to exchange knowledge and information on the views of different stakeholders about the link between national and EU level policy framework for Rare Diseases, and to raise awareness around the collaborative efforts needed to highlight clear policy recommendations for National and EU level framework.

The meeting addressed key topics, such as access to clinical trials, integrated care, innovative treatments and the need for a European Action Plan. View the full meeting programme.

THE MAIN OPPORTUNITIES MENTIONED BY THE PARTICIPANTS WERE:

- NPRD updated and integrated in the National Strategy for Health 2023- 2030 with funding
- PNRR – Funds from the National Program for Recovery and Resilience has objectives that support care for RDs
- POS – Funds from the National Operational program for Health – funding for Connection of European activities and Genetic Centers
- POIDS - Operational Programme Social Inclusion and Dignity
- Good collaboration between RONARD and other stakeholders involved in Romanian care ecosystem
- EU collaboration with Eurordis and ERNs, including participation in JARDIN

THE MAIN CHALLENGES IDENTIFIED IN THE CARE SYSTEM IN ROMANIA WERE:

- No national registry for people living with a rare disease.
- No ORPHAcode implemented (just ICD10 which doesn't cover most of the diagnosis).
- No proper care infrastructure.
- No financial support for Connection of European activities or PO.
- Protracted waiting time between the approval of an Orphan Drug by the EMA and the drug being made accessible for patients in Romania.
- No national program for genetic testing for adults with RDs.
- Not always good communication between medical and social services, and other stakeholders.

SATELLITE MEETING
Organised by RONARD

WE HAVE TO ACT TOGETHER TO:

- Maintain or improve our collaboration in to achieve as many objectives of the NPRDs as possible through PNRR, POS and POIDS.
- Monitor annually the implementation of the NPRD through our Europlan/ national conference.
- Ensure that our objectives can be found in the local strategies, national strategy for health and care but also in the recommendations and European Action Plan for Rare Diseases

NEXT STEPS

1. To organise a meeting at the Ministry of Health to discuss the Operational Plan for 2024 of NPRD.
2. Follow the 3 objectives presented by the Ministry of Health as priorities for RDs in 2024: upscaling of NBS, RD national registry and National Coordination Hub and present the results at the RD School for Journalists 2024 and Europlan conference.
3. Organise actions for upscaling of NBS: Webinar on 25th June in Bucharest.
4. Share Experience webinars to promote our objectives.
5. Continue to work on rare disease law.

THOUGHT LEADER SESSIONS

HARNESSING DIGITAL SOLUTIONS FOR RARE DISEASES

25 APRIL 2024, 12.00-13.15 CET

OVERVIEW

This ECRD 2024 Thought Leader Session, chaired by Jelena Malinina, highlighted the vital role of digital solutions in tackling challenges for individuals with rare diseases. Experts from various fields shared innovations, discussed policy impacts, and explored patient perspectives on digital health tools.

SPEAKERS

- Jelena Malinina, EURORDIS Data Director
- Bruno Sarfati, CEO and Founder, Elise de Beauvais
- Dr. Tudor Groza
- William Declerck, M.D.
- Prof. Lizbeth Goodman, Director, SMARTlab/UCD
- Huw Williams, CTO/Co-director, SMARTlab
- Irina Kubinschi, MedTech Europe, Manager Government Affairs & Public Policy
- Daniel Lewi, The Cure & Action for Tay-Sachs (CATS) Foundation

FEATURED DIGITAL TOOLS

ASWEKNOW

A tool streamlining the diagnostic process, providing reliable information, and linking to specialised care, thus enhancing the overall management and understanding of rare conditions.

UTOPIA CASE STUDY

An example of using computational models to understand disease progression and provide valuable insights for patients and clinicians.

MINA

A chatbot aiming to improve patient-doctor communication with AI-powered Q&A platform.

KEY DISCUSSION POINTS

- Enhanced personalisation and engagement: digital tools should cater to the unique needs of rare disease patients, ensuring meaningful engagement.
- Data Protection and ethical concerns: maintaining high standards of data security and addressing ethical concerns, especially with AI-driven tools, is crucial.
- Policy support for innovation: supportive policies are needed to foster innovation and facilitate patient access to technologies.
- Engagement with policymakers: Stakeholders should engage with policymakers to shape regulatory environments.
- Collaboration across sectors: continuous collaboration among technology developers, healthcare professionals, patients, and policymakers is essential.
- Ongoing evaluation and feedback: continuous real-world evaluation and feedback from end-users are vital for development.

KEY CONCLUSIONS

To successfully develop digital tools for rare diseases, the following actions are needed:

- Enhanced communicational knowledge: widespread knowledge sharing to inform all stakeholders.
- Analysing phenotype relationships: understanding relationships between phenotypes to improve tool accuracy.
- Interoperability rules and policies: clear rules promoting seamless data exchange among health systems and digital tools.
- Collaborative development: Pooling resources and expertise among researchers, developers, patients, and healthcare professionals.
- Common Free Application Programming Interfaces (APIs): providing free APIs to access knowledge bases and data repositories, fostering innovation.
- European Health Data Space: leveraging high-quality data for research to advance digital solutions.
- Rigorous data protection measures: Implementing stringent protocols to safeguard patient information, complying with GDPR.
- Ensuring access and equity: ensuring digital solutions reach all population segments, especially underserved regions, to prevent healthcare disparities.

COLLABORATING FOR CHANGE: TRANSFORMING RARE DISEASE OUTCOMES THROUGH PUBLIC-PRIVATE PARTNERSHIPS

3 MAY 2024, 15.00 - 16.00 CET

CO-POWERED BY

RARE DISEASE MOONSHOT
SCALING UP PUBLIC-PRIVATE PARTNERSHIPS TO ACCELERATE RESEARCH IN RARE DISEASES

Together4RD
RARE DISEASES

PARTICIPANTS

Attendees included representatives from the European Commission, patient groups and advocates, researchers/European Reference Networks and industry.

SPEAKERS

- Magda Chlebus (EFPIA and Rare Disease Moonshot),
- Sheela Upadhyaya (Together4RD),
- Danielle Dong (Sanofi),
- Kira Gillett (The Bespoke Gene Therapy Consortium),
- Roseline Favresse and Matt Bolz-Johnson (EURORDIS),
- Alexandre Bétourné (Critical Path Institute),
- Salah-Dine Chibout (Novartis),
- Holm Graessner (ERN-RND)

MAIN TOPICS OF DISCUSSION

The panel discussion focused on the transformative potential of public-private partnerships (PPPs) in rare disease research and therapy development. They emphasised the critical role PPPs play in fostering innovation, particularly through integrating genomics into routine healthcare for precision-based treatments. Despite substantial investments, 95% of rare diseases still lack effective treatments, highlighting the need for stronger partnerships. The session explored the importance of co-creation, emphasising transparent communication, mutual goals, and early integration of diverse perspectives. Standardising data collection methods and creating centralised platforms were identified as pivotal for facilitating more efficient regulatory processes and therapy development. Pre-competitive collaboration was highlighted as a significant opportunity to enhance the collaborative spirit within PPPs. The primary challenges discussed included the complexity of rare disease research, the necessity for robust partnerships, effective data governance, and overcoming political and operational obstacles. The opportunities lie in leveraging these partnerships to accelerate research, enhance patient outcomes, and drive innovation and efficiency in developing new treatments.

MAIN CONCLUSIONS

Strengthening PPPs is essential for advancing rare disease research and therapy development. Enhanced collaboration among industry, academia, government agencies, and patient organisations is crucial. Integrating genomics into routine healthcare enables precision-based treatments and improves patient outcomes. Moreover, the effective co-creation, transparent communication, and mutual goals are vital for successful partnerships. Standardising data collection and creating centralised platforms streamline regulatory processes and accelerate new therapies access. Finally, maintaining a pre-competitive environment fosters collaboration and addresses the intricate challenges in rare disease research. These conclusions highlight the importance of a multi-faceted approach to drive innovation and efficiency, ultimately improving patient outcomes.

NEXT STEPS

Industry leaders and research institutions must advocate for new partnership models and work to remove political and operational barriers. Patient organisations should engage actively in these collaborations to ensure patient needs are central to all initiatives.

Researchers (public and private) and research funders should focus on achieving long-term goals through step-by-step advancements. Continuous adjustments and improvements based on ongoing findings are essential for developing more robust and effective outcomes.

Continue the efforts to sustain Public - Private Partnerships in the pre-competitive area while ensuring the deployment of outcomes in real-world settings.

PPP coordinators should establish robust frameworks with clear guidelines for roles, responsibilities, and communication channels to facilitate seamless coordination and decision-making within partnerships and ensure efficiency.

THE GLOBAL COMMISSION TO END THE DIAGNOSTIC ODYSSEY FOR CHILDREN WITH A RARE DISEASE

7 MAY 2024, 14.00 - 15.00 CET

A multi-disciplinary group of rare disease advocates, researchers, physicians, and innovators from across the globe with a commitment to shortening the long and arduous diagnostic journey. More than 50 participants joined the session, including clinicians, patient groups and advocates, researchers and industry partners.

GLOBAL COMMISSION
to End the Diagnostic Odyssey for Children with a Rare Disease

Main topics of discussion, challenges and opportunities: Two of the Global Commission's co-chairs re-introduced the Global Commission and set the stage for the panel discussion on accelerating diagnosis.

Neil Inhaber, Vice President and Head of Global Medical Functions and Rare Diseases at Takeda, described the Global Commission's vision: a clear path to a timely, accurate diagnosis for all children.

Yann Le Cam, co-founder of EURORDIS, presented how the Global Commission's focus aligns with global health policy priorities and provided an overview of a new framework that the Global Commission created to guide and support countries in their efforts to improve and accelerate diagnosis of children with a rare disease.

THE GLOBAL COMMISSION'S FRAMEWORK CONSISTS OF THREE KEY PILLARS:

Timed with ECRD, the Global Commission launched a report with the new framework and case studies associated with each pillar, highlighting success stories in efforts to accelerate diagnosis.

A panel discussion with Global Commission members then focused on the importance of spotlighting exemplar programmes and sharing best practices on diagnosis so that countries can learn from each other.

The panel was moderated by **Mike Porath**, patient advocate and founder and CEO of The Mighty, and included:

- **Gareth Baynam**, a practicing clinical geneticist at the Rare Care Centre at Perth Children's Hospital in Western Australia, Director of the Undiagnosed Diseases Program and Founder of Lyfe Languages, universal Indigenous medical translator initiative
- **Roberto Giugliani**, the director and practicing clinical geneticist at Casa dos Raros, a specialised rare disease centre in Porto Alegre Brazil
- **Alexandra Heumber Perry**, CEO of Rare Diseases International

The panellists emphasized the value of highlighting examples of initiatives, such as Casa dos Raros (an integrated diagnostic and treatment center in Brazil), that have been able to reduce the time to diagnose a rare disease, noting the importance of addressing cultural and policy-related barriers to diagnosis.

The session concluded with co-chair **Alaa Hamed**, Global Head of Medical Affairs Rare Diseases at Sanofi, reinforcing why the Global Commission's work is so important given that the average time to receive an accurate rare disease diagnosis is still 4-5 years.

MAIN CONCLUSIONS

Regrettably, access to diagnosis depends on where a patient lives and the efficiency or sophistication of their healthcare system – however, there are emerging innovations that can help leapfrog these challenges. When looking to scale diagnostic efforts across different contexts, interventions should consider unique barriers affecting target populations.

NEXT STEPS

The Global Commission is seeking examples of innovative and compelling work to accelerate diagnosis that it can turn into case studies for dissemination to the rare disease community. Please email suggestions to info@globalrarediseasecommission.com.

POSTERS

CLICK HERE TO VIEW THE

POSTER SHOWCASE

1ST PLACE

32 | ERN-EYE

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

Caroline Wernert-Iberg

ADhoc – An immersive Serious Game

Raising awareness among healthcare professionals about announcing a diagnosis of Rare Diseases

Authors : [1&2] Caroline WERNERT-IBERG, [1] Dorothée LEROUX, [2] Marilyne OSWALD, [3] Dr Pascal DUREAU, [3] Dr Catherine VIGNAL-CLERMONT, [4] Dr Catherine VINCENT-DELORME, [5] Dr Elise SCHAEFER, [6] Russell WHEELER, [1,2 & 5] Pr Hélène DOLLFUS

[1] ERN-EYE Network, Hôpitaux Universitaires de Strasbourg, France [2] Filière SENSGENE, Hôpitaux Universitaires de Strasbourg, France
[3] Hôpital Fondation Adolphe de Rothschild, Paris, France [4] CHRU de Lille, France [5] Hôpitaux Universitaires de Strasbourg, France [6] ERN-EYE Supporting Partner

Introduction

The French rare diseases network SENSGENE has created an innovative and didactic Serious Game with the aim of raising awareness among healthcare professionals of all ages and specialities about best practices when announcing a diagnosis of rare diseases. **The European Reference Network ERN-EYE translated into English.**

Facing a surprised or disoriented patient, calming a father's anger, or managing unruly siblings, all while delivering a complex diagnosis - these are the objectives drawn from real-life situations that players are confronted with.

Issues

Announcing a diagnosis is an aspect of a doctor's profession that is often delicate and not necessarily part of their training.

The serious game called "ADhoc" aims to remind players of good practice and allow them to immerse themselves in real-life situations. The game allows players to test their expertise and measures their progress by means of "good practice" and "empathy" gauges, which fluctuate as players answer questions.

Goals of the Serious Game

A fast Serious Game that can be played anywhere!

ADhoc's main objective? To learn where you want, when you want and quickly!

In the role of a doctor in 15 short scenes separated into 3 levels of difficulty, the player enhances their expertise and measures progress through the "best practices" and "empathy" gauges, which fluctuate based on responses. At the end of each scene, the player can review the key pedagogical concepts to remember.

15 DIFFERENT SCENES

3 LEVELS OF DIFFICULTY

1 HOUR TO PLAY

Materials and Methods

Developed by the French company Almédia, this serious game was scripted during focus groups by a committee of medical experts from the SENSGENE and ERN-EYE networks, including Pr Hélène Dollfus, coordinator of both networks. Patient associations and psychologists were also consulted to validate the content.

The Committee of medical experts was composed of Pr Hélène Dollfus, SENSGENE and ERN-EYE coordinator, Dr Pascal Dureau, Adolphe de Rothschild Foundation Hospital, Paris, Dr Elise Schaefer, Strasbourg University Hospitals, Dr Catherine Vignal-Clermont, Adolphe de Rothschild Foundation Hospital, Paris, and Dr CatherineVincent-Delorme, Lille University Hospital. The serious game was translated and edited with the help of Russell Wheeler, supporting partner of ERN-EYE.

To make the content fun and original, SENSGENE turned to Almédia (www.almedia.fr) and asked Metapraxis (https://metapraxis.fr/) to act as assistant project manager.

Results

Diffused since April 2023, more than 800 players (rare diseases healthcare professionals) logged into the game, with more than a third playing all 15 scenes offered. This Serious Game has been a success since its launch with a rating of 4.8/5. Here are some feedbacks:

"This game is a great reminder of what not to do."

"I spent two evenings 'playing' and drew some improvements for my practice, as well as several avenues for reflection"

The Serious Game can be accessed on computers, tablets and smartphones using the QR code below or at: https://prod08.almedia.fr/erneye/

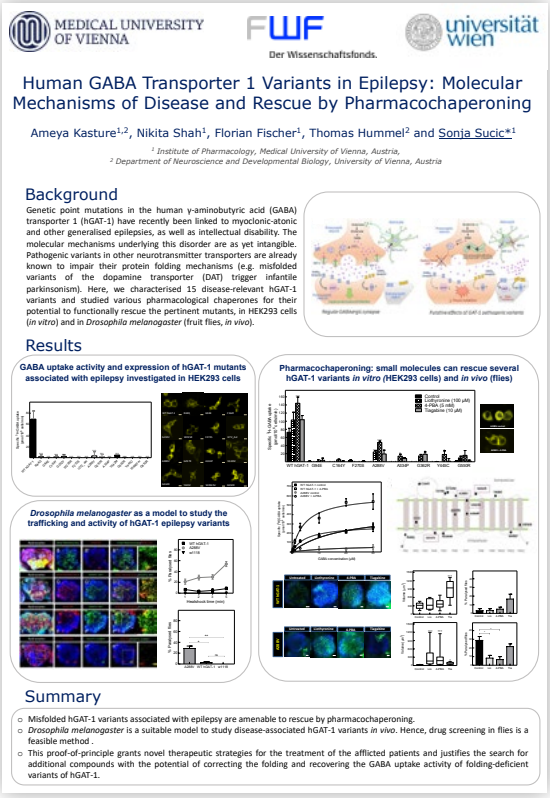
SCAN ME

RUNNER UP

7 | Medical University of Vienna

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

Sonja Sucic



MEDICAL UNIVERSITY OF VIENNA · FWF Der Wissenschaftsfonds. · universität wien

Human GABA Transporter 1 Variants in Epilepsy: Molecular Mechanisms of Disease and Rescue by Pharmacochaperoning

Ameya Kasture[1,2], Nikita Shah[1], Florian Fischer[1], Thomas Hummel[2] and Sonja Sucic*[1]

[1] Institute of Pharmacology, Medical University of Vienna, Austria,
[2] Department of Neuroscience and Developmental Biology, University of Vienna, Austria

Background

Genetic point mutations in the human γ-aminobutyric acid (GABA) transporter 1 (hGAT-1) have recently been linked to myoclonic-atonic and other generalised epilepsies, as well as intellectual disability. The molecular mechanisms underlying this disorder are as yet intangible. Pathogenic variants in other neurotransmitter transporters are already known to impair their protein folding mechanisms (e.g. misfolded variants of the dopamine transporter (DAT) trigger infantile parkinsonism). Here, we characterised 15 disease-relevant hGAT-1 variants and studied various pharmacological chaperones for their potential to functionally rescue the pertinent mutants, in HEK293 cells (*in vitro*) and in *Drosophila melanogaster* (fruit flies, *in vivo*).

Results

GABA uptake activity and expression of hGAT-1 mutants associated with epilepsy investigated in HEK293 cells

Pharmacochaperoning: small molecules can rescue several hGAT-1 variants *in vitro* (HEK293 cells) and *in vivo* (flies)

***Drosophila melanogaster* as a model to study the trafficking and activity of hGAT-1 epilepsy variants**

Summary

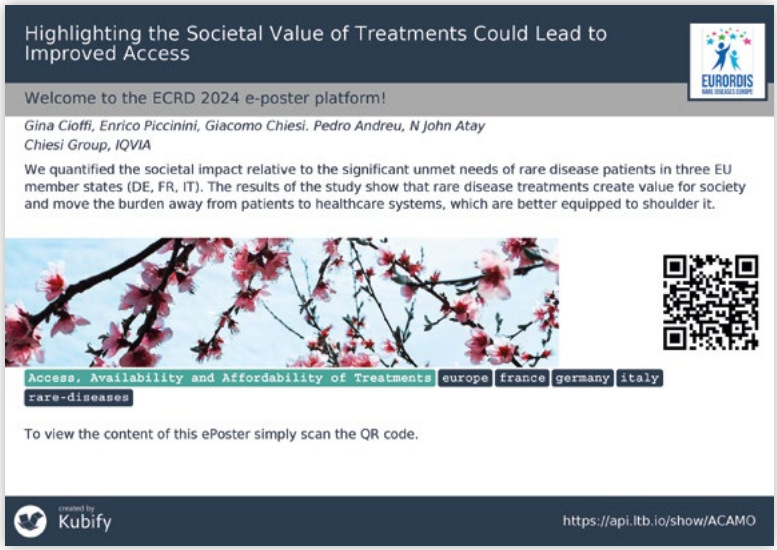
- Misfolded hGAT-1 variants associated with epilepsy are amenable to rescue by pharmacochaperoning.
- *Drosophila melanogaster* is a suitable model to study disease-associated hGAT-1 variants *in vivo*. Hence, drug screening in flies is a feasible method .
- This proof-of-principle grants novel therapeutic strategies for the treatment of the afflicted patients and justifies the search for additional compounds with the potential of correcting the folding and recovering the GABA uptake activity of folding-deficient variants of hGAT-1.

RUNNER UP

98 | Chiesi Global Rare Diseases

Highlighting the Societal Value of Treatments could Lead to Improved Access

Gina Cioffi



Highlighting the Societal Value of Treatments Could Lead to Improved Access

EURORDIS RARE DISEASES EUROPE

Welcome to the ECRD 2024 e-poster platform!

Gina Cioffi, Enrico Piccinini, Giacomo Chiesi. Pedro Andreu, N John Atay
Chiesi Group, IQVIA

We quantified the societal impact relative to the significant unmet needs of rare disease patients in three EU member states (DE, FR, IT). The results of the study show that rare disease treatments create value for society and move the burden away from patients to healthcare systems, which are better equipped to shoulder it.

Access, Availability and Affordability of Treatments | europe | france | germany | italy | rare-diseases

To view the content of this ePoster simply scan the QR code.

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ERN GENTURIS
With every diagnosis we can help an entire family

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SOCIAL MEDIA

Manchester Rare Conditions Centre @mft_IMRare · May 15

Great to see that Eva, our Engagement Lead, is attending the European Conference on Rare Diseases.The rare disease community is constantly growing stronger through worldwide connection, collaboration,patient advocacy & policy change. Excited to hear what Eva learns at #ECRD2024

1 · 9 · 277

EJP RD - European Joint Programme on Rare D @EJPRareDise: · May 15

Check out the #ECRD2024 comic strip showcasing major milestones from the 1st ECRD to today and beyond! ⭐ At labor #7, we celebrate the launch of EJP RD in 2019, marking a major step forward in unifying and accelerating rare disease research

download2.eurordis.org/ecrd/2024/ecrd...

Michelle Muscat • 2nd

National Alliance For Rare Diseases Support Malta & The Mari...

1mo • + Follow

It is time Malta takes this seriously. Whereas we know how well treated patients in Malta are, it is time that a National Action Plan for Malta on rare diseases is drafted, budgeted for and eventually implemented. Rare disease patients and their families need to find a system that ensures they get the best approaches, the best treatments, the best support services as any other patient with any other disease. The European Reference Networks have to start working for Maltese patients too and we need policy makers that believe in this and not just pretend they are interested. Soon enough there will be a European Action Plan. We still need to see the realties of our small island state, but I am sure that with a good will, emphaty and of course determination we will provide an equal and equitable future to our rare patients. Jo Etienne Abela EURORDIS-Rare Diseases Europe #ecrd2024

Matt Bolz-Johnson and 57 others · 1 repost

raising rareness (Lyndsey Walsh) @raisingrareness · May 15

Attending the 2024 European Conference on Rare Diseases & Orphan Products online 🙌

#ECRD2024 #ActRare #RareCommunity #CareForRare #IndividuallyRareCollectivelyCommon #30MillionReasons

@eurordis @ERNithaca

Orphanet @Orphanet · May 24

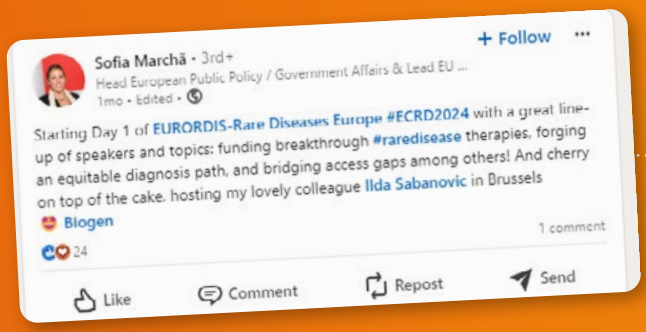
Sign the #ECRD2024 Open Letter! ✍

Join EURORDIS and the many other signatories in addressing the future leaders of Europe and calling on them to address the urgent needs of the rare disease population. #ActRare2024

Read the full letter and sign now! go.eurordis.org/mm8ssb


JWMDRC @jwmdrc · May 23
Sign the #ECRD2024 Open Letter!
Join @eurordis and the many other signatories in addressing the future leaders of Europe and calling on them to address the urgent needs of the rare disease population. #ActRare2024
Read the full letter and sign now! go.eurordis.org/mm8ssb
SIGN THE OPEN LETTER
15 & 16 MAY 2024


HYBRID
ACTION WITHIN REACH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Starting Day 1 of EURORDIS-Rare Diseases Europe #ECRD2024 with a great line-up of speakers and topics: funding breakthrough #raredisease therapies, forging an equitable diagnosis path, and bridging access gaps among others! And cherry on top of the cake, hosting my lovely colleague Ilda Sabanovic in Brussels
Biogen
1 comment
24
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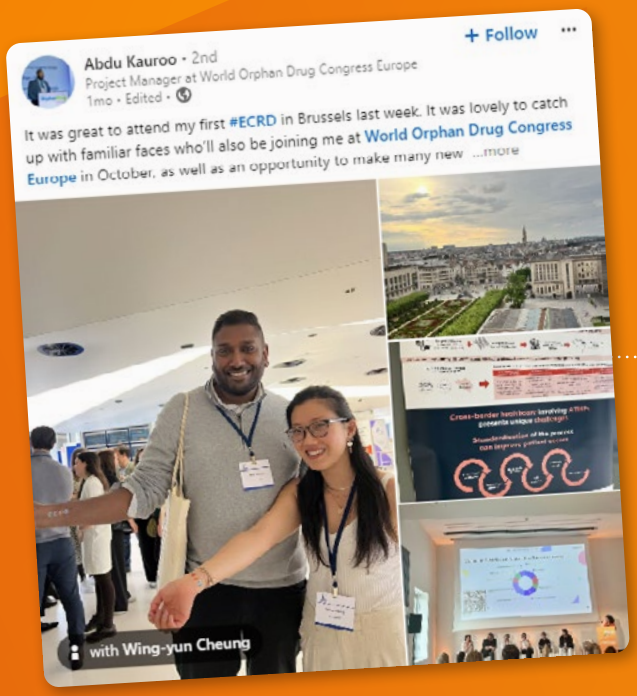
Fundacja Matio @fundacjamatio · May 16
Trochę w Krakowie trochę w Brukseli, ale jak coroku na ECRD. Jak zwykle, merytoryczne spotkanie społeczności Chorób Rzadkich. #ECRD2024, @eurordis , @yann_eurordis , @fundacjamatio @PaweWjtowicz12 @chorobyrzadkie
ACTION WITHIN REACH
15 16 MAY 2024
3
59


Melissa Kinch @Melissa_Kinch_ · May 15
Excited to help co-create a #MentallyHealthy toolkit for #RareDiseases
Great to see such amazing things like this come out of #ECRD2024
NO HEALTH WITHOUT MENTAL HEALTH!
EURORDIS-Rare Diseases Europe


Avril Daly @avrilbdaly · May 15
Welcome Vicky's little helper! #ECRD2024 #rarediseases
Rare Disease Policy @RareDiseasesEU · May 15
Great to be back in #Brussels for #ECRD2024 (with a little helper this time)..@avrilbdaly & new @eurordis CEO @VirginieBF31 emphasise the importance of this conference for shaping policy for #raredisease
WHAT IS ECRD?
8
171


Abdu Kauroo • 2nd
Project Manager at World Orphan Drug Congress Europe
1mo • Edited •
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It was great to attend my first #ECRD in Brussels last week. It was lovely to catch up with familiar faces who'll also be joining me at World Orphan Drug Congress Europe in October, as well as an opportunity to make many new ...more
with Wing-yun Cheung


Flaminia Macchia • 2nd
Head, Global Rare Disease Policy
1mo • Edited •
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All stakeholders signing the Open Letter calling for a European Action Plan on Rare Diseases #ActRare2024
#ECRD2024
Eva Schoeters Daria Julkowska Oliver Sude Ana Rath @alain coheur Avril Daly Tina Taube Filipa Júlio Virginie Bros-Facer Holm Graessner
Matt Bolz-Johnson and 154 others
1 comment • 5 reposts


ERN EuroBloodNet @ERNEuroBloodNet · May 15
Check the @ERNEuroBloodNet posters at #ECRD2024!
ERN-EuroBloodNet facilitating access to cross-border healthcare for patients with rare hematological diseases in the European Union
The role of ENROL Registry on Advancing Translational and Clinical Research in Hematology
European Commission and 7 others
4
99


ACTION
WITHIN REACH
ERKNet


Genetic Alliance UK @GeneticAll_UK · May 16
Genetic Alliance UK are at #ECRD2024 presenting different posters. Come and find our Head of Policy and Engagement Manager for Scotland, Natalie to learn the findings of a recent healthcare professionals survey undertaken in Scotland.
2
12
375


Daniel de Vicente @Danidevic · May 15
Comienza el #ECRD2024
Reconocido como el mayor evento mundial liderado por pacientes de #enfermedadesraras con un dialogo abierto y colaborativo entre pacientes, Administraciones, investigadores, clínicos e industria farmacéutica.
3
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501

HYBRID ecrd

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PIONEERING SOLUTIONS FOR RARE DISEASES

HYBRID 15 & 16 MAY 2024

12th European Conference on Rare Diseases and Orphan Products

MEDIA PARTNERS

beacon for rare diseases

DDN EXPLORING DRUG DISCOVERY AND DEVELOPMENT

EMJ

ean european academy of neurology

gms

RDODJ Rare Disease and Orphan Drugs Journal

Raconteur

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