

HYBRID



3 - 4 JUNE 2026 • PRAGUE & ONLINE

13<sup>th</sup> EUROPEAN CONFERENCE ON RARE DISEASES AND ORPHAN PRODUCTS

# RARE DISEASES IN A CHANGING & COMPETITIVE EUROPE

SHAPING POLICIES TO ADDRESS THE UNMET NEEDS  
OF PEOPLE LIVING WITH RARE DISEASES

## EXECUTIVE SUMMARY



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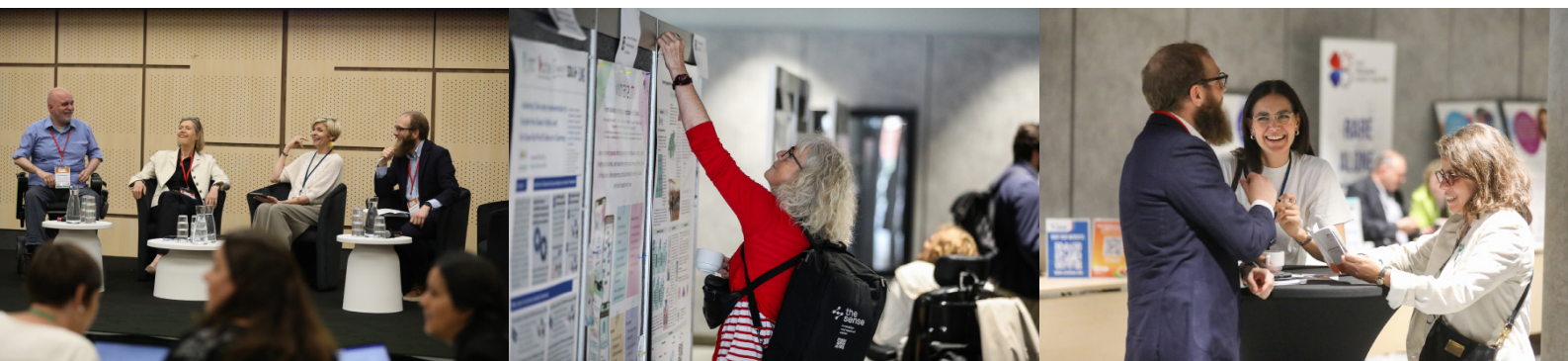


#ECRD2026

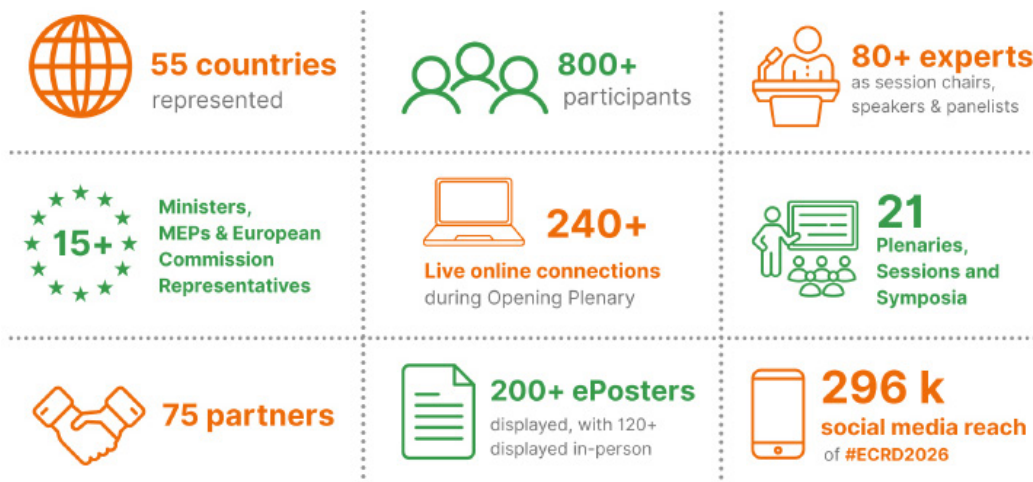
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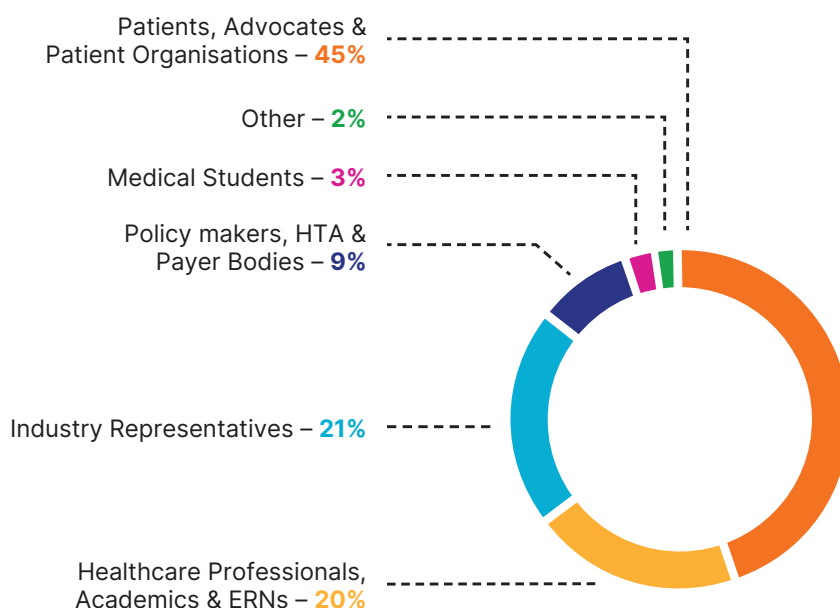


# KEY FACTS AND FIGURES



As Europe's largest patient-led rare disease conference, the European Conference on Rare Diseases and Orphan Products provides a unique forum for dialogue, learning and collaboration across the rare disease community. The conference brings together diverse stakeholders to exchange ideas, build consensus and help shape rare disease policy at the European level.

The 13th edition of the European Conference on Rare Diseases and Orphan Products (ECRD) took place on 3–4 June 2026, welcoming over 500 participants to the O2 universum Congress Center in Prague, alongside more than 200 online attendees. The conference's hybrid format enabled experts, advocates, policymakers, researchers and industry representatives from 55 countries to connect, exchange perspectives and foster collaboration across the rare disease community. Together, participants explored policy priorities and practical solutions to improve the lives of people living with a rare disease and their families. With the help of our custom-built virtual 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 80 expert speakers and chairs. Throughout the conference, attendees contributed to interactive networking activities, participated in small-group discussions, and shared research and best practices through a hybrid showcase of more than 200



# SETTING THE STAGE

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Five years after the launch of **Rare2030** and midway towards the **2028 WHO Global Plan target**, ECRD 2026 took place at a pivotal moment for rare disease policy in Europe. Following the deployment of Europe's Life Sciences Strategy and in the wake of the adoption of the Biotech Act, the conference highlighted how investment in rare disease research is not only essential to improving outcomes for patients, but also to strengthening Europe's competitiveness in biotechnology, precision medicine and life sciences innovation.

Under the overarching title of **Rare Diseases in a Changing & Competitive Europe: Shaping Policies to Address the Unmet Needs of People Living with Rare Diseases**, ECRD 2026 explored the key opportunities and challenges shaping the future of the rare disease ecosystem. The programme addressed a wide range of policy priorities, including therapy development and access, timely diagnosis, holistic and specialised care, health technology assessment, and mental health.

ECRD 2026 was the second fully hybrid edition of the conference, connecting hundreds of online participants with those attending in person in Prague through a highly interactive virtual platform. This format broadened participation, bringing together voices from a wide range of countries, backgrounds and perspectives, and enriching discussions across the rare disease community.

ECRD 2026 marked a key milestone in the inclusive, multi-stakeholder process leading to the development of a **European Blueprint for Rare Diseases**. Building on discussions held during pre-conference meetings, a dedicated workshop and throughout the conference programme, the Blueprint will be published in Autumn 2026, providing a shared framework for action, driving lasting impact across Europe while reinforcing Europe's contribution to the WHO Global Action Plan on Rare Diseases.

By participating in ECRD 2026, attendees played an active role in shaping the future of rare disease policy in Europe and beyond - helping to drive real, lasting change for millions of people living with a rare disease.



# KEYNOTE SPEECHES

## OPENING PLENARY



**Avril Daly, President, EURORDIS – Rare Diseases Europe**

“ECRD really is about patients. It’s about the families. It’s also about clinicians and researchers and policymakers and regulators and advocates and industry partners. We are all united by [...] a shared commitment to improving the lives of people living with rare diseases.”



**Jean Saslawsky, Chief Executive Officer, EURORDIS – Rare Diseases Europe**

“I have a deep connection with the mission of EURORDIS and a great admiration also for all the work that has been achieved for almost three decades by the rare disease community in Europe.”



**Olivér Várhelyi, European Commissioner for Health and Animal Welfare**

“We need to keep working together to improve the lives of people with rare diseases and their families, and you can continue to count on the Commission.”



**Adam Vojtech, Minister for Health, Czech Republic**

“The call for a comprehensive European Action Plan for Rare Diseases remains highly relevant. It continues to resonate strongly across the community of patients, families, and stakeholders who rightly expect that Europe will provide a more coordinated, predictable, and equitable framework of care.”



**Prof. Leonidas Phylactou, CEO and Medical Director, Cyprus Institute on Neurology and Genetics**

“All Europeans should have an equal access to good hospitals, good treatment places, good medicines. Currently, this is not the case. That’s why the pan-European policy will help towards that.”



**Daniel Naumovas, Vice Minister of Health, Lithuania**

“Even in times of crisis, we still need to push forward innovation. And what better showcase of an innovative country if not the case that you can treat all patients?”



**Kamil Šaško, Minister for Health, Slovakia**

“Real progress in my eyes means much better involvement of patients, of patient organisations, well-educated patient organisations who can really advocate, who can bring arguments.”



**Javier Padilla Bernáldez, Secretary of State for Health, Spain**

“We cannot be thinking about co-designing or about engaging patient organisations in the process of the development of the interventions without guaranteeing the financial stability and the financial sustainability for these organisations.”



**Vytenis Andriukaitis, Member of the European Parliament, Lithuania**

“Ultimately, the question is not whether Europe knows what needs to be done. We do. The question is whether we are prepared to organise ourselves differently and act with greater ambition.”



**Tomasz Dylag, Deputy Head of Unit, Unit D2 Health Innovations & Ecosystems, DG Research & Innovation, European Commission**

“We need to turn research into clinical reality and into real world impact. We know that collaboration isn’t optional, it’s the only way forward. That means in practice that we need to have policymakers, clinicians, researchers, companies working together more.”

# KEYNOTE SPEECHES



**Noah Higón Bellver, Jurist, Political Scientist, Activist and Writer**

"I want a Europe where receiving a diagnosis doesn't depend on luck, a Europe where research is not a privilege, a Europe where access to treatment doesn't come too late, a Europe that understands that care is not an expense, it is a form of justice."



**Marcin Nowacki, President, TEN Section – European Economic and Social Committee (EESC)**

"Rare diseases show that competitiveness and social progress are not competing objectives. They can and should reinforce one another."



**Nathalie Moll, Director General, European Federation of Pharmaceutical Industries and Associations**

"There is a real momentum today, a genuine opportunity to push the boundaries, to create the right conditions to have deeper collaboration and accelerate the development of solutions for patients who can't wait."



**Ana Rath, Director, Orphanet**

"Let's work towards an integrated European Rare Disease Plan that delivers. In this conference, we will work and have your ideas, your collective intelligence towards this goal."

## CLOSING PLENARY



**Enrique Terol, Health Counsellor, Permanent Representation of Spain to the EU**

"One message has come through very clearly. European ambitions must be translated into practical tools that improve the lives of patients and support healthcare systems. Europe can provide directions and coordination, but implementation at the end happens in member states."



**Vicky McGrath, Chief Executive Officer, Rare Diseases Ireland**

"We need to drive this change. And it's not just for the benefit of our health, it's for the benefit of Europe's health, and it's for the benefit of innovation and the future."



**Holm Graessner, Managing Director – Rare Disease Centre, University Hospital Tübingen and HLM4RARE**

"The conclusion is simple. We have built the networks. The next task is to enable them to become innovation networks so that our expertise does not only serve to these patients, but also generate tomorrow's evidence, diagnostics, and treatment."



**Stine Bosse, Member of the European Parliament, Denmark**

"All this has to be for all patients. We really also need to work together to make sure we don't hold each other back, but we find methods of funding so that we get this done [...] for all patients across the European Union."



**Kateřina Konečná, Member of the European Parliament, Czech Republic**

"What we need now is not another resolution. We need a binding, adequately funded EU Action Plan for Rare Diseases, one that sets clear timelines, measurable targets, and real accountability mechanisms."

# KEYNOTE SPEECHES

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**Nicolás Gonzáles Casares, Member of the European Parliament, Spain**

"We need more. We have to be ambitious. We have an opportunity now to raise this issue very strongly and say to the people that we need a plan."



**Elena Nevado del Campo, Member of the European Parliament, Spain**

"Europe has already shown that when there is a clear strategy, common goals and adequate funding, it can change patients' lives [...] Now we must do the same with rare diseases."



**Antonio Parenti, DG Santé, European Commission**

"For decades, patient advocates have been the driving force behind the progress in rare diseases, and they have transformed rare diseases from what has been often an invisible condition into a recognised European public health priority"



**Natasha Azzopardi-Muscat, Director of Health Systems, WHO Regional Office for Europe**

"The EURORDIS European Blueprint, developed together with Rare Diseases International and a broad multi-stakeholder community, is going to be a really important contribution toward the future European Action Plan and towards shaping Europe's input and experience into the Global Action Plan itself."



**Alain Coheur, President, CCMI, European Economic and Social Committee (EESC)**

"It's occasions like this, organised by EURORDIS, where like-minded advocates come together, where we take the time to exchange, to reconnect, and where the seeds of the new project are planted"



**Alexander Natz, Secretary General, European Confederation of Pharmaceutical Entrepreneurs (EUCOPE)**

"I'm really optimistic, because of conferences like ECRD, that we can get things done together in collaboration"



**Maria del Mar Manu Pereira, Scientific Director, ERN-EuroBloodNet**

"The next decade must be about ensuring that every person living with a rare disease can really access that expertise, treatment, and timely diagnosis."



**Jean Saslawsky, Chief Executive Officer, EURORDIS – Rare Diseases Europe**

"EURORDIS has always been much more than an advocacy organisation. It's a movement, a movement of patients and families living with rare diseases."



**Avril Daly, President, EURORDIS – Rare Diseases Europe**

"From Prague, we leave with a very clear message. The momentum is here. The community is united. The tools and the knowledge exist. What we must now match them with is sustained political will, coordination, and investment in that action. We will not stop until every person living with a rare disease in Europe can access the diagnosis, care, and treatment that they deserve."

# EUROPEAN BLUEPRINT FOR RARE DISEASES:

## PRE-CONFERENCE WORKSHOP

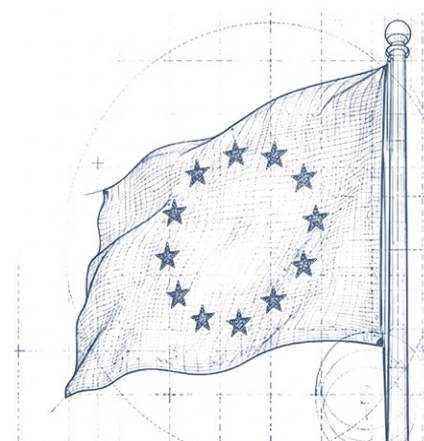
The **Pre-ECRD Workshop on the European Blueprint for Rare Diseases**, held on **2 June 2026** in Prague ahead of the European Conference on Rare Diseases and Orphan Products (ECRD 2026), received more than 150 registrations from across Europe, including patient representatives, policymakers, clinicians, researchers, industry, regulators and international organisations. The workshop represented a key milestone in the development of the European Blueprint for Rare Diseases, providing an opportunity to validate, refine and prioritise the proposed policy actions that will underpin both a future **EU Action Plan on Rare Diseases** and **contribute to the development of a Global Rare Disease Action Plan**.

The workshop combined plenary sessions with six facilitated thematic breakout discussions, each focused on a core pillar of the Blueprint. Participants assessed proposed policy actions against a common set of prioritisation criteria, considering their potential impact, ability to address unmet needs and inequities, feasibility, and capacity to enable progress across other areas. Alongside identifying priority actions, discussion groups proposed amendments, refinements and opportunities to merge related actions, ensuring that the emerging Blueprint reflects both strategic ambition and practical implementation.

The six discussion groups examined complementary dimensions of rare disease policy:

- ▶ **Integrated and person-centred care and social inclusion**, focusing on multidisciplinary care pathways, psychosocial support, social protection and meaningful involvement of people living with rare diseases.
- ▶ **Research and innovation**, addressing sustainable investment, interoperable research infrastructures, patient partnership and innovative approaches to research and clinical development.
- ▶ **Access to treatments and medical devices**, exploring equitable access, regulatory and health technology assessment (HTA) innovation, cross-border collaboration and sustainable reimbursement pathways.
- ▶ **Earlier, faster and more accurate diagnosis**, examining newborn screening, structured diagnostic pathways, professional education, genomic medicine and support for undiagnosed patients.
- ▶ **Access to specialised and highly specialised healthcare**, considering the integration and long-term sustainability of European Reference Networks (ERNs), national rare disease plans and coordinated care pathways.
- ▶ **Optimising the rare disease data ecosystem**, focusing on interoperability, implementation of OrphaCodes, the European Health Data Space (EHDS), registries, governance and responsible use of health data.

Together, these discussions provided a comprehensive review of the proposed Blueprint, enabling participants to identify the actions considered most critical at both European and global levels while generating practical recommendations to strengthen the coherence, implementation and long-term impact of the future policy framework.



# WORKSHOP CONCLUSIONS

Across all six thematic discussions, a strong consensus emerged that the challenges facing people living with rare diseases require a more coordinated, integrated and ambitious policy response. Although each discussion group identified distinct priorities, participants consistently recognised that **progress depends on strengthening the enabling conditions that connect diagnosis, care, research, treatment access and data systems**, rather than pursuing isolated interventions.

Several cross-cutting themes were evident throughout the workshop. Participants emphasised the importance of **integrated, person-centred care**; meaningful **involvement of patients and families in policy, research and service design**; stronger **collaboration** between European and national stakeholders; **sustainable investment** in infrastructures and expertise; and **interoperable data systems** capable of supporting research, clinical care and evidence-based policymaking. Across multiple discussions, participants also highlighted the need to **reduce fragmentation** by improving coordination between health, social care, research and regulatory systems, while promoting greater **consistency across Member States**.

The workshop also demonstrated broad support for prioritising actions that create lasting systemic capacity. These included strengthening **multidisciplinary care pathways**, reinforcing **research and clinical infrastructures**, modernising **regulatory and access frameworks**, improving **diagnostic pathways**, embedding **European Reference Networks** more effectively within national health systems, and developing a robust and interoperable rare disease **data ecosystem**. Collectively, participants viewed these foundational measures as essential for accelerating innovation, reducing inequalities and improving outcomes for people living with rare diseases across Europe and beyond.

Overall, the workshop confirmed broad stakeholder support for a dedicated **European Action Plan on Rare Diseases** capable of translating shared priorities into coordinated action. The discussions reinforced that many rare disease challenges transcend national borders and require sustained European and **international collaboration** to ensure equitable access to diagnosis, care, research, treatments and data-driven innovation. The outcomes of the workshop will directly inform the finalisation of the European Blueprint for Rare Diseases, alongside wider consultation with the Core Group, Regional Task Force and broader stakeholder community, before publication of the final Blueprint in October 2026.



# PROGRAMME COMMITTEE

## CHAIRS



**Magda Chlebus**  
EFPIA



**Pavla Doležalová**  
General University  
Hospital Prague



**Vicky McGrath**  
Rare Diseases  
Ireland



**Enrique Terol  
Garcia**  
Permanent  
Representation of  
Spain to the EU

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**Valentina Bottarelli**  
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**Annalisa Scopinaro**  
UNIAMO



**Jan Swiderski**  
Kyowa Kirin International

# PARALLEL SESSIONS

## DAY 1

### THERAPIES & MEDICAL DEVICES, DEVELOPMENT & ACCESS



#### SESSION 1: Shaping the Future of Rare Disease Therapies: Innovation, Trials and Regulation in Europe's New Biotech Era

**MODERATOR** ▶ **Tim Leest**, Chair of the European Committee for Orphan Medicinal Products

**SPEAKERS** ▶ **Dr Daria Julkowska**, Scientific Coordinator, ERDERA  
▶ **Victor Maertens**, EUCOPE  
▶ **Dr Alexandre Méjat**, AFM - Téléthon  
▶ **Samantha Parker**, Italfarmaco

#### SESSION 2: Medical Devices for People Living with a Rare Disease

**MODERATOR** ▶ **Dr Tomek Grybek**, Foundation of Borys the Hero, EURORDIS Board Member

**SPEAKERS** ▶ **Dr Donal O'Connor**, HPRA and EC task forces on orphan devices and breakthrough technology  
▶ **Dr Kolaleh Eskandarian**, KPI  
▶ **Dr Aleksandra Krygiel-Nael**, Johnson & Johnson  
▶ **Dr Anneliene Jonker**, Duchenne Parent Project and leader of DeCODE project  
▶ **Dr Andrea Rappagliosi**, HTA Committee, MedTech Europe

### DIAGNOSIS, RESEARCH AND PREVENTION



#### SESSION 1: Early Detection & Newborn Screening (NBS): Feasibility, Cost and Equity

**MODERATOR** ▶ **Martina Cornel, M.D., PhD**, European Society of Human Genetics

**SPEAKERS** ▶ **Dani Bancroft**, Genetic Alliance UK  
▶ **Sofia Douzgou Houge, M.D., PhD, FRCP**, Haukeland University Hospital, and Norwegian Centre for Rare Diagnoses  
▶ **Prof. Janbernd Kirschner**, Screen4Care  
▶ **Simon Lande**, Health Lumen  
▶ **Dr Laurent Pasquier**, University Hospital Centre Rennes

#### SESSION 2: The Journey from Rare Disease Diagnosis to Treatment

**MODERATOR** ▶ **Dr Gulcin Gumus**, EURORDIS - Rare Diseases Europe

**SPEAKERS** ▶ **Prof. Urh Groselj**, University Medical Centre Ljubljana  
▶ **Dr Kelly Ormond**, ETH-Zürich and Stanford University  
▶ **Dr Andreas Øberg**, Oslo University Hospital  
▶ **Dr Mariette Driessens**, VSOP

**PRESENTERS** ▶ **Prof. Jean Bergounioux**, Raymond Poincaré Hospital, APHP  
▶ **Dr Ana Rath and Robin Sarfati**, Rare Disease Knowledge (RDk)

## EVIDENCE-BASED HOLISTIC CARE



### SESSION 1: Advancing Holistic Care for Rare Conditions: A Patient-Centred and Evidence-Based Approach

**MODERATOR** ▶ **Claudia Crocione**, HHT Europe

**SPEAKERS** ▶ **Jessie Dubief**, EURORDIS - Rare Diseases Europe  
▶ **Iolo Eilian**, HSE Ireland  
▶ **Dr Roser Francisco**, Hospital Sant Joan de Déu  
▶ **Arne Noppe**, Sciensano  
▶ **Dr Rohita Sharma**, Alexion

### SESSION 2: Filling the Gaps to Provide Evidence-Based Holistic Care

**MODERATOR** ▶ **Jessie Dubief**, EURORDIS - Rare Diseases Europe

**SPEAKERS** ▶ **Prof. David Keegan**, ERN-EYE  
▶ **Dr Pauline Nauroy**, Fondation Maladies Rares  
▶ **Dr Solange Roumengous**, APHP - BNDMR France  
▶ **Eva-Maria Strömsholm**, Finnish Association of Gynaecological Cancer Patients  
▶ **Dr Natalie Uhlenbusch**, University Hospital Hamburg - Q.RARE.LI Project

## DAY 2

## SPECIALISED HEALTHCARE



### SESSION 1: Strategies to Improve Access to Specialised Healthcare for People Living with a Rare Disease

**MODERATOR** ▶ **Vicky Hedley**, Newcastle University

**SPEAKERS** ▶ **Anna Arellanesová**, Rare Diseases Czech Republic  
▶ **Cecilia Gunnarsson, M.D.**, Linköping University  
▶ **Prof Luca Lovrečić**, University Medical Centre Ljubljana  
▶ **Dr Holm Graessner**, University Hospital Tübingen and HLM4RARE

### SESSION 2: Rethinking Access to Specialised Healthcare for PLWRD – What Should We Measure?

**MODERATORS** ▶ **Dr Enrique Terol**, Permanent Representation of Spain to the EU  
▶ **Ines Hernando**, EURORDIS - Rare Diseases Europe

**SPEAKERS** ▶ **Dr Maria Karanikolos**, European Observatory on Health Systems and Policies  
▶ **Charline Maertens**, KCE Belgian Health Care Knowledge Centre  
▶ **Natalie Frankish**, Head of Public Affairs and Lead for Scotland, Genetic Alliance UK  
▶ **Dr Anne-Sophie Lapointe**, Ministry of Solidarity and Health, France

## PREPARING REIMBURSEMENT DECISIONS



### SESSION 1: A State of the Art of the HTA Regulation (HTAR)

**MODERATOR** ▶ **François Houÿez**, EURORDIS – Rare Diseases Europe

**SPEAKERS** ▶ **Dr Stephanie Said**, HTA Coordination Group, Subgroup on JSC, Federal Joint Committee, Gemeinsamer Bundesausschuss (G-BA)  
▶ **Camelia Isaic**, HAE Junior  
▶ **Maciej Gajewski**, Ipsen

### SESSION 2: Preparing Reimbursement Decisions - Practical Consequences and Perspectives on the Future

**MODERATOR** ▶ **Julien Delaye**, Brussels Center for Collaboration in Health (BCCH)

**SPEAKERS** ▶ **Dr Roisin Adams**, National Centre for Pharmacoeconomics, Ireland  
▶ **François Houÿez**, EURORDIS – Rare Diseases Europe  
▶ **Matteo Scarabelli**, EFPIA  
▶ **Gaetan Duport**, European Haemophilia Consortium  
▶ **Dr Jose Valverde**, DG Santé, European Commission

## MENTAL HEALTH



### SESSION 1: Rethinking Mental Health in Rare Conditions: From Undefined Challenges to Collaborative Solutions

**MODERATOR** ▶ **Dr Kirsten Johnson**, Fragile X International

**SPEAKERS** ▶ **Isabella Brambilla**, Dravet-Italia, CREA & EpiCARE  
▶ **Jessie Dubief**, EURORDIS - Rare Diseases Europe  
▶ **Vinciane Quoidbach**, European Brain Council  
▶ **Dr Stewart Rust**, Manchester University NHSFT  
▶ **Dr Jane Waite**, Cerebra Network



## SHAPING THE FUTURE OF RARE DISEASE THERAPIES: INNOVATION, TRIALS AND REGULATION IN EUROPE'S NEW BIOTECH ERA

This session examined how scientific innovation, regulatory reform and cross-sector collaboration can accelerate the development of therapies for people living with rare diseases. While advances in biotechnology are creating new opportunities, speakers agreed that significant barriers remain, particularly for conditions affecting very small populations; for these small populations one key challenge lies in determining what level of evidence is appropriate for regulatory approval. Participants recognised that conventional evidence requirements are often difficult to achieve in ultra-rare diseases, raising important questions about how regulators can balance timely access to innovative therapies with the need for safety and effectiveness.

Non-profit organisations remain key drivers of rare disease innovation yet are not adequately recognised within current European funding and policy frameworks. There is a need for funding models that support clinical development as well as research, alongside regulatory pathways and compliance requirements that are better adapted to the realities of ultra-rare disease development. Whilst existing regulatory tools could improve sustainability if used more consistently, wider acceptance of real-world evidence would help address evidence gaps, particularly in paediatric neurodegenerative diseases (as brought up during the panel discussion). The proposed European Biotech Act offers unique opportunities to address these challenges and may be able to build on established EU infrastructures such as ERDERA and the European Reference Networks, while strengthening data interoperability, education and capacity building.

Despite cross-border initiatives such as ERDERA and the ERNs, Europe's fragmented landscape continues to create barriers to therapy development, regulation and access. While there is broad agreement on what needs to change, the challenge now lies in moving from ideas to implementation. Achieving this will require stronger coordination across stakeholders, with a shared vision from the outset and a clearer focus on the desired outcomes. This is exactly the type of coordination sought by EURORDIS in its advocacy work towards an EU Action Plan for Rare Diseases (see Blueprint work).

Finally, the discussion highlighted the importance of embedding patient perspectives far earlier in therapy development. Speakers argued that people living with ultra-rare diseases should play a greater role in shaping clinical trial design, selecting meaningful endpoints and informing risk-benefit assessments. While patient involvement is increasingly recognised, participants agreed that stronger methodologies are needed to ensure these perspectives consistently inform regulatory decision-making. They also stressed that regulatory considerations should be integrated from the earliest stages of development rather than addressed only later in the process.

Accelerating therapy development for ultra-rare diseases will require more flexible regulatory approaches, greater acceptance of innovative evidence generation, sustainable funding for diverse developers, and stronger patient partnership throughout the development pathway. These principles offer valuable lessons for building a more collaborative and patient-centred healthcare innovation ecosystem.





## MEDICAL DEVICES FOR PEOPLE LIVING WITH A RARE DISEASE

Medical devices are playing a growing role in improving the lives of people living with rare diseases, not only in the diagnosis and treatment of disease but also in improving independence and quality of life and, in many cases, becoming an essential part of care. This session examined how regulation, innovation and new approaches to evidence generation can better support their development, focusing on creating a regulatory environment that better reflects the realities of rare disease innovation while ensuring patients can benefit more quickly from new technologies.

Many medical devices are highly customised and must adapt to patients' changing needs – particularly in paediatric care – therefore necessitating development pathways that recognise continuous innovation rather than one-off products, whether the device is a diagnostic tool, a life-sustaining technology, a personalised implant or an assistive device.

However, current regulatory frameworks remain largely designed for common conditions and larger patient populations. Conventional evidence requirements are often difficult to meet in rare diseases, where randomised clinical trials may be impractical or even unethical. Recent European guidance allowing greater flexibility was welcomed, though speakers agreed that more proportionate regulatory pathways, clearer methodological guidance and stronger incentives are still needed. Several participants called for a dedicated orphan medical device framework to encourage innovation for small patient populations.

Emerging tools – real-world evidence, patient registries, artificial intelligence, digital twins and in silico trials – can complement traditional clinical studies and generate evidence where conventional trials are not feasible. Participants noted that these approaches could accelerate access while supporting ongoing evidence generation after market approval, provided robust governance and data standards are in place.

Throughout the session, speakers emphasised that devices should be designed around outcomes that matter most to people living with rare diseases: including independence, participation and quality of life. Greater involvement of patients and caregivers throughout development – from identifying unmet needs to defining meaningful endpoints and generating evidence – is seen as essential to producing technologies that deliver real-world benefit.

Finally, the panel recognised that regulatory approval alone does not guarantee access. Reimbursement, procurement and fragmented healthcare pathways continue to limit the availability of many innovative devices. Participants concluded that accelerating medical device innovation will require proportionate regulation, innovative evidence-generation methods, targeted incentives for developers and stronger collaboration across the rare disease ecosystem, ensuring that medical devices are fully recognised as a core component of patient-centred rare disease care.





## EARLY DETECTION & NEWBORN SCREENING (NBS): FEASIBILITY, COST AND EQUITY

This session examined the opportunities and challenges of genetic newborn screening (gNBS) in improving the early identification of rare diseases. Whilst advances in genomic technologies are expanding the potential of newborn screening, there is a continued and unmet need for robust care pathways, equitable implementation and greater collaboration across Europe.

A central discussion concerned the development of clear frameworks to decide which conditions should be included in genetic newborn screening programmes. While prevalence remains an important consideration, speakers agreed that penetrance and clinical actionability are equally critical when determining whether a condition is suitable for screening. Participants argued that future approaches should move beyond assessing individual diseases in isolation towards transparent criteria that can evolve alongside scientific knowledge. There was also support for developing a common European framework with a minimum list of conditions, while recognising that implementation remains the responsibility of individual Member States.

As highlighted during the discussion, successful screening programmes depend on much more than identifying a genetic variant. Comprehensive care pathways, which include confirmatory diagnosis, genetic counselling, treatment where available, long-term monitoring and psychosocial support for families, are essential. As sequencing costs fall, speakers noted that the greater challenge lies not in the screening itself but in ensuring that healthcare systems have the capacity to provide appropriate follow-up care.

With minority populations often underrepresented in genomic databases and biobanks, participants highlighted the need for equity, as such underrepresentation can reduce the accuracy of genetic interpretation and limit the benefits of genomic medicine for some communities. Improving diversity in research, strengthening public engagement and ensuring clear, accessible information for families were identified as important steps towards more equitable implementation.



The session also addressed the needs of people who remain undiagnosed despite genomic testing: a group that should be recognised as distinct within the rare disease community and supported through dedicated care pathways and research initiatives. Advances in genomic medicine should therefore not only increase diagnosis rates but also strengthen support for those whose conditions remain unexplained.

The wider societal and ethical implications of expanded newborn screening were also explored, including the psychological impact of identifying conditions that may not develop until later in life or may never become clinically significant. The session concluded that the future of genetic newborn screening will depend on balancing scientific innovation with patient-centred implementation. This will need to be supported by shared European frameworks, inclusive data, sustainable care pathways and meaningful engagement with patients and families at every stage of the screening process.



## THE JOURNEY FROM RARE DISEASE DIAGNOSIS TO TREATMENT

Advances in newborn screening, artificial intelligence (AI) and collaborative diagnostic approaches are transforming the early identification of rare diseases, from metachromatic leukodystrophy (MLD) to spinal muscular atrophy (SMA). Through practical examples and emerging initiatives, speakers demonstrated how innovation can shorten the diagnostic odyssey and improve access to timely care, while stressing that advances in diagnosis are matched by equitable access to treatment and coordinated care pathways.

Speakers highlighted that effective advanced treatment depends on identifying affected children before symptoms appear, making newborn screening essential for delivering approved therapies at the right time. Therapy development and newborn screening should not be viewed as separate processes, but as complementary elements of a single patient pathway. Close collaboration between clinicians, researchers, policymakers and decision-makers was identified as key to implementing screening programmes that can serve as models for other rare diseases.

Discussions also raised the persistent inequalities affecting access to diagnosis, clinical trials and innovative treatments. While scientific progress is creating new therapeutic opportunities, participants noted that high costs and uneven access continue to prevent many patients from benefiting. Addressing these disparities will require greater coordination across healthcare systems to ensure that earlier diagnosis translates into earlier intervention.

The session also explored the essential role of genetic counselling and ethical decision-making in supporting patients beyond diagnosis. While often associated with testing and diagnosis, genetic counselling plays an equally important role throughout the treatment pathway, helping patients and families navigate uncertainty around treatment effectiveness, long-term outcomes, participation in

clinical trials, and the unknown risks and benefits of emerging gene therapies. While advances in gene and genome editing offer unprecedented hope, access remains uneven across Europe due to disparities in healthcare systems, geography, expertise, and availability of genetic counselling services. The need for continued emotional, informational, and practical support, including connecting families with appropriate resources and support networks was also highlighted.

Artificial intelligence was presented as a potential tool to complement existing diagnostic approaches. AI-based analysis of motor function, for example, is already being used to identify children with SMA at an earlier stage. Rather than replacing newborn screening, speakers discussed AI's potential to refine and strengthen existing programmes by supporting earlier recognition of symptoms and helping identify children who may otherwise be missed.

The session also explored broader efforts to reduce the diagnostic odyssey across the rare disease community. New digital tools are being developed to support diagnosis across multiple rare diseases, while participants highlighted the importance of recognising undiagnosed patients as a distinct group with specific needs. Speakers called for care pathways that extend beyond diagnosis, bringing together multidisciplinary expertise, digital resources and stronger links with European Reference Networks to support patients whose conditions remain unidentified.

The session concluded that earlier diagnosis depends not only on technological innovation but also on integrated, patient-centred systems of care. Combining newborn screening, AI-supported diagnosis, multidisciplinary collaboration and equitable access to treatment offers an opportunity to improve outcomes across the rare disease community while ensuring that no patient is left behind, including those who remain undiagnosed.



## ADVANCING HOLISTIC CARE FOR RARE CONDITIONS: A PATIENT-CENTRED AND EVIDENCE-BASED APPROACH

High-quality patient-generated evidence can significantly strengthen rare disease policy, improve healthcare services and support more meaningful patient partnership. Yet whilst there is growing recognition of patient-centred approaches across the rare disease ecosystem, speakers in this session emphasised that lived experience alone is not sufficient to drive change. Instead, patient perspectives must be combined with robust data and effective policy design in order to influence decision-making and create sustainable improvements in care.

The discussion began by examining the role of patient surveys in generating evidence. Speakers stressed that individual surveys should not be viewed in isolation but as one component of a wider evidence base, complementing registries, published research and population-level data. In the rare disease context, demonstrating that survey or registry participants are fully representative of the wider patient population remains inherently impossible, given the limited knowledge on the prevalence of individual rare diseases and the lack of knowledge about the overall demographics of people living with rare diseases. Rather than weakening their value, participants argued that surveys provide unique insights into patient experiences that are often absent from other data sources. By complementing clinical registries, survey data can identify patients who have not accessed specialist services or Centres of Expertise, providing a more complete understanding of unmet needs and inequalities in access to care.

The session highlighted the importance of quantifying the broader burden of rare diseases. Findings from recent economic analyses demonstrated the substantial health, social and financial impacts experienced by patients and families, including frequent misdiagnosis, reduced quality of life and significant societal costs. Current estimates put the economic burden of rare diseases at around €250 billion per year across Europe, with 78 days of productivity lost per year across each person living with a rare disease and their caregivers. Participants therefore welcomed ongoing work to develop standardised methods for measuring disease burden across Europe using Disability-Adjusted Life Years (DALYs), which could provide policymakers with more consistent evidence to inform resource allocation and health policy.



Speakers also explored how patient-reported experience measures (PREMs) can support continuous quality improvement within healthcare services. Examples from clinical practice demonstrated how combining validated patient experience tools with qualitative feedback from patients and families enables hospitals to identify areas for improvement and embed patient perspectives directly into service design and organisational decision-making.

The session concluded by examining patient partnership at a system level, using a case study of Ireland's evolving model of patient involvement to illustrate how partnership can be embedded across healthcare, research, policy and governance through dedicated structures, formal training and sustainable investment. Meaningful patient engagement, the participants agreed, requires more than consultation: it depends on building the evidence, skills and organisational culture needed for patients to become equal partners in shaping healthcare systems.



## FILLING THE GAPS TO PROVIDE EVIDENCE-BASED HOLISTIC CARE

This session discussed how rare disease research can better support holistic, patient-centred care by expanding beyond traditional clinical outcomes to include social, economic and psychosocial dimensions. Holistic care is currently defined and evaluated in an inconsistent and unstandardised manner, with concepts such as integrated, person-centred and holistic care often used interchangeably despite describing a wide range of interventions. This lack of standardisation makes it difficult to compare studies, identify best practices and scale successful approaches across different healthcare systems.

Alongside definitional inconsistency, current research continues to prioritise short-term clinical outcomes, while paying less attention to quality of life, wellbeing and other outcomes that matter most to patients and families. Speakers argued that future research should be designed around patients' everyday lives rather than healthcare settings alone. This includes involving patients as equal partners throughout the research process, from defining meaningful outcomes to interpreting findings; and studying the interaction between health, social and environmental factors, while ensuring that research produces practical recommendations applicable in daily life.

The discussion highlighted the growing importance of national and European rare disease registries in supporting research, service planning and policymaking. Examples from France's National Database for Rare Diseases and the developing ERN-EYE REDgistry illustrated how harmonised, interoperable registries can facilitate natural history studies, epidemiological research, trial feasibility assessments and secondary data reuse. Participants stressed that common data standards and FAIR principles are essential for enabling meaningful collaboration across countries and improving the use of real-world data.

As discussed in this session, the social sciences and humanities also play an important role in addressing unmet needs that extend beyond biomedical research. Despite this, participants noted that psychosocial care, quality of life and broader societal impacts remain underrepresented within rare disease research and funding. Expanding interdisciplinary collaboration, creating dedicated funding opportunities and strengthening education for researchers and policymakers were identified as important steps towards addressing these gaps.

Delivering genuinely holistic care requires a stronger evidence base that reflects the full realities of living with a rare disease. Combining patient partnership, multidisciplinary research, interoperable data infrastructures and greater investment in social as well as biomedical research may allow the rare disease community to develop more effective, scalable and patient-centred models of care across Europe.





## STRATEGIES TO IMPROVE ACCESS TO SPECIALISED HEALTHCARE FOR PEOPLE LIVING WITH A RARE DISEASE

Recognising that no single model can meet the needs of people living with rare diseases in every country, this session drew on examples from Sweden, the Czech Republic, Slovenia and Germany to explore practical approaches to improving access to specialised healthcare in a diverse range of contexts. Through these European examples, speakers examined how national generic rare disease care pathways, cross-border healthcare, European Reference Networks (ERNs) and National Reference Networks can work together to ensure that patients access the right expertise at the right time while keeping care close to home where possible.

The discussion opened by highlighting the priorities identified by the rare disease community, including access to specialist care, effective care coordination and better support in navigating often complex healthcare systems. Participants agreed that achieving these goals requires stronger alignment between local, national and European initiatives, supported by improved data sharing and policy integration.

Sweden's experience demonstrated the value of a generic care pathway applicable across a range of rare and complex conditions, regardless of diagnosis. Developed in partnership with patient organisations, the pathway aims to shorten the time to diagnosis, strengthen multidisciplinary coordination and ensure that every patient has an individual care plan and a designated care coordinator. National implementation is supported by shared indicators and patient-reported data while allowing each region to adapt the pathway to local circumstances. Participants also recognised the importance of engaging primary care professionals through dedicated education and training.

Experience from the Czech Republic illustrated how collaboration between patient organisations and Cross-Border Healthcare National Contact Points can help families navigate administrative requirements, reimbursement procedures and access to specialist centres abroad. Participants emphasised that patients often identify appropriate expertise themselves and called for greater recognition of specialist clinical judgement when decisions about cross-border care are made.

Examples from Slovenia and Germany demonstrated how national ERN Coordination Hubs and national reference networks can connect national expertise with European collaboration, facilitating knowledge exchange, professional networking and wider dissemination of best practice. At the same time, participants acknowledged the resource constraints faced by many countries and stressed that sustainable funding and administrative support will be essential for these structures to reach their full potential.

Speakers agreed that improving access to specialised healthcare requires coordinated action across all levels of the healthcare system. While ERNs provide expertise, knowledge and collaboration, national structures remain essential for translating these resources into routine patient care. Strengthening care pathways, supporting cross-border collaboration and investing in well-coordinated national networks will be key to delivering more equitable access to specialised healthcare for people living with rare diseases across Europe.





## RETHINKING ACCESS TO SPECIALISED HEALTHCARE FOR PLWRD - WHAT SHOULD WE MEASURE?

Timely access to specialised healthcare is widely recognised as a major challenge for people living with a rare disease – yet countries currently measure it in a fragmented way, making it difficult for policymakers to get a full picture and identify where improvements are most needed. This session focused on developing a practical set of indicators that can help policymakers understand not only whether specialised services exist, but whether patients can actually reach and benefit from them.

While some countries monitor different aspects relevant to access, such as waiting times to first appointment at a Centre of Expertise or equity of access, participants identified significant gaps. Access to cross-border healthcare remains largely absent from existing monitoring frameworks, despite its importance for patients requiring highly specialised expertise. Other gaps included limited measurement of care pathways and continuity of care, workforce capacity and linking access to specialist services gaps with health outcomes and patient experience.

Speakers demonstrated how Health System Performance Assessment (HSPA) frameworks can provide a more structured approach to evaluating access to specialist rare disease care. By examining governance, financing, workforce, service delivery and health outcomes together, these frameworks help identify the system-level factors that influence access, quality and equity. Adapting HSPA to the rare disease context would enable countries to compare performance over time, identify policy gaps and better understand the impact of healthcare reforms.

The discussion also highlighted the NEED initiative, which seeks to measure healthcare from the perspective of unmet patient needs rather than health system structures alone.

By developing standardised indicators covering access, quality of care, treatment burden, research and treatment effectiveness, the initiative aims to ensure that future policies reflect the priorities of people living with rare diseases and support more equitable access to innovation.

Throughout the session, speakers emphasised that meaningful indicators must remain patient-centred. Speakers broadly supported measuring elements such as access to a designated care coordinator, individual care plans, multidisciplinary care, successful transitions between services and holistic assessment of physical, psychological and social needs. The session also highlighted the importance of developing patient-reported experience measures (PREMs) and patient-reported outcome measures (PROMs) specifically for rare diseases, recognising that patients themselves are best placed to evaluate whether healthcare systems are meeting their needs.

Looking to the future, a shared European framework of practical, patient-centred indicators could strengthen national rare disease plans, support more consistent policy evaluation and help identify inequalities that remain hidden within healthcare systems. Together, these measures could provide policymakers with the evidence needed to improve access, quality and equity for people living with rare diseases across Europe.



# TRACK E, SESSION 1



## A STATE OF THE ART OF THE HTA REGULATION (HTAR)

In light of the recent implementation of the European Health Technology Assessment Regulation, this session explored how the new framework can best support more consistent evidence generation across Europe while ensuring that assessments better serve the needs of national decision-makers (pricing and reimbursement decisions). This session paid particular attention to Joint Scientific Consultations (JSCs), Joint Clinical Assessments (JCAs) and the meaningful involvement of patients throughout these processes.

Participants discussed the early implementation of Joint Scientific Consultations, noting that demand currently exceeds available capacity. However, speakers expressed confidence that the number of available consultations will increase as the new system develops, allowing more developers to benefit from early scientific advice and improved alignment between regulators and HTA bodies.

Meaningful involvement and patient participation, speakers agreed, require more than simply inviting patient representatives to participate. Patient experts need appropriate preparation, training and sufficient time to engage effectively, alongside the confidence to contribute to technical discussions and an understanding of conflict-of-interest requirements. Existing training initiatives were highlighted as valuable resources for strengthening patient capacity and ensuring that patient representatives can participate as equal partners within the HTA process.

The discussion also focused on the types of evidence considered within Joint Clinical Assessments. Participants emphasised that assessments should capture the full impact of a disease and its treatment by considering not only clinical outcomes but also patient-reported outcome measures (PROMs), quality of life and caregiver burden. These outcomes provide important insights into the real-world value of therapies and are particularly relevant for rare diseases, where improvements in daily functioning and quality of life may be as meaningful as traditional clinical endpoints.



Speakers acknowledged that integrating patient-centred evidence into HTA remains an evolving area. PROMs collected prospectively during clinical trials are currently easier to incorporate than data generated through routine clinical practice or other real-world settings. Participants therefore highlighted the need for stronger methodologies and validation standards to improve the consistency and acceptance of quality-of-life and caregiver burden data within future assessments. European Reference Networks were identified as important partners in supporting the generation, standardisation and validation of these data.

The new European HTA framework therefore offers an important opportunity to strengthen patient-centred decision-making across Europe. However, achieving this ambition will require continued investment in methodological development, patient training and collaboration between HTA bodies, researchers, clinicians, European Reference Networks and patient organisations to ensure that future assessments reflect the outcomes that matter most to people living with rare diseases and their families.

# TRACK E, SESSION 2



## PREPARING REIMBURSEMENT DECISIONS - PRACTICAL CONSEQUENCES AND PERSPECTIVES ON THE FUTURE

Where the first session of this track examined the mechanics of the new HTAR, this session asked what it means in practice – focusing on reimbursement decisions, operational realities, and the path ahead for Joint Clinical Assessments (JCAs) and Joint Scientific Consultations (JSCs).

Rather than a finished system, the current framework should be viewed as a learning process. Although implementation is already underway, procedures continue to be refined through real-world experience rather than formal pilot projects. Speakers expressed confidence that the framework will become more efficient and streamlined over time as stakeholders gain practical experience and identify opportunities for improvement.

Speakers discussed the importance of coordination and shared understanding across the different actors involved in HTA. Developers, regulators, HTA bodies and other stakeholders need to align expectations as early as possible in the development process, particularly regarding evidence requirements and assessment methodologies. Greater consistency and flexibility were seen as essential to ensuring that the new framework supports timely and predictable assessments while responding to the specific challenges of rare diseases.

The discussion also highlighted the operational challenges associated with implementing the Regulation. The European Commission continues to play an important coordinating role by supporting Member States and facilitating the operation of the new system. However, speakers recognised that resource and capacity constraints remain

significant as the number of Joint Clinical Assessments and Joint Scientific Consultations continues to increase. Addressing these challenges will be essential to maintaining the quality and sustainability of the framework as it expands. Patient involvement emerged as another central theme, with participants agreeing that patients should contribute not only to individual assessments but also to the ongoing development of the framework itself. Meaningful engagement was seen as an opportunity to strengthen dialogue between patients, clinicians, regulators, HTA bodies and national authorities while ensuring that assessments better reflect unmet needs and the realities of living with a rare disease. Speakers also highlighted the importance of clearly defining patient communities and establishing structured mechanisms for patient participation throughout the assessment process. The Commission reminded all that this is the first EU Regulation that organises the systematic involvement of patients in all procedures.

Whilst the European HTA Regulation represents an important milestone in strengthening collaboration across Europe, implementation will require continued refinement, adequate resources and close coordination between stakeholders. Nonetheless, participants agreed that the framework provides a strong foundation for more consistent, transparent and patient-centred health technology assessments. Ensuring that all stakeholders work towards common objectives and that patient perspectives remain embedded throughout implementation will be key to the framework's long-term success.



# TRACK F, SESSION 1



## RETHINKING MENTAL HEALTH IN RARE CONDITIONS: FROM UNDEFINED CHALLENGES TO COLLABORATIVE SOLUTIONS

Drawing on new evidence, such as from the Rare Barometer survey, alongside clinical experience and patient perspectives, this session explored the multiple factors that shape mental health for people living with rare diseases and their families.

Findings from the Rare Barometer survey showed that seven in ten respondents reported poor mental health, with symptoms of anxiety and depression, but also loneliness and suicidal thoughts occurring at rates far higher than in the general population. Many participants experienced multiple mental health challenges simultaneously, while half received no support during their diagnostic journey. Those who did receive support often reached a diagnosis more quickly, illustrating the importance of psychological and social support alongside medical care. Despite high levels of need, professional mental health services remain difficult to access, with cost and availability continuing to create significant barriers.

The discussion also emphasised that rare diseases affect entire families, not only individual patients. Parents and caregivers, particularly mothers, often experience substantial emotional and practical burdens, while family relationships may be affected by prolonged uncertainty, intensive care needs and financial pressures. Supporting family resilience should therefore be recognised as an essential component of rare disease care, alongside greater attention to bereavement support and the needs of siblings and caregivers.

Another emerging theme was the need to embed mental health support across the entire life course. Participants highlighted significant gaps during key transitions, particularly the move from paediatric to adult services, when many young people and families experience disruption in care and increased isolation. Examples of good practice, including transition coordinators and integrated multidisciplinary teams, demonstrated how continuity of care can improve both clinical and psychosocial outcomes.



The discussion also explored practical ways to strengthen mental healthcare within rare disease services. Speakers called for routine mental health screening, greater training for healthcare professionals, psychologists, nurses and other frontline staff, and stronger integration of mental health professionals within multidisciplinary teams. Participants also highlighted the importance of involving young people directly in service design, recognising that their perspectives should complement those of parents and caregivers.

A key takeaway from this session was that mental health should be recognised as a core component of rare disease care rather than an optional addition. Participants called for an ambitious European approach that embeds psychological support throughout the care pathway, strengthens family resilience, improves access to specialised services and expands research into the long-term mental health impacts of rare diseases.

# GROUP DISCUSSIONS

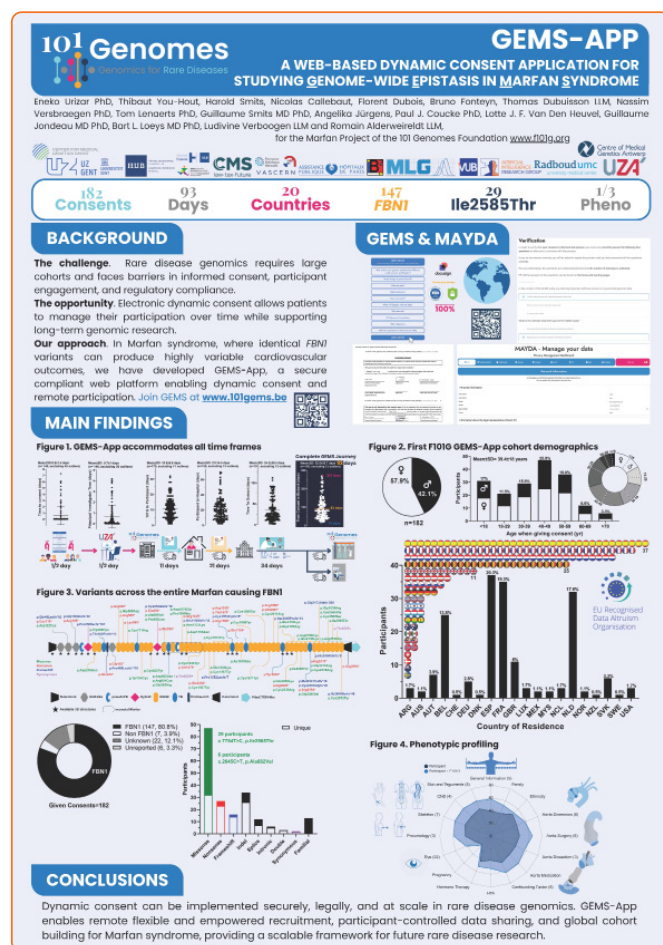
This ECRD, in-person attendees were invited to propose topics around which to moderate small-group discussions. These took place as part of a dedicated networking session and covered a broad range of topics and approaches, across the six tracks of the conference. From therapeutic session on the challenges of patient advocacy, to exploring how to better design PROMs, and a roundtable on Digital Health Technologies, these group discussions created a meaningful and interactive space for collaboration and the exchange of experience and ideas between participants.

The 11 selected groups were titled and moderated as follows:

- ▶ **The Weight of the Whisper: What Advocates Carry** – moderated by Vesna Aleksovska
- ▶ **Targeted incentives for the development of therapies for ultra-rare conditions: what could the EU do?** – moderated by Rosa Castro
- ▶ **How can we truly capture what ‘treatment success’ means for patients? The powerful complementarity between patient-reported outcomes (PROs) and digital health technologies (DHTs)** – moderated by Céline Desvignes-Gleizes
- ▶ **When diagnosis fails: rare conditions beyond classification, data and research systems** – moderated by Roseline Favresse
- ▶ **Addressing Systemic Barriers to Rare Children’s Access to Medicines** – moderated by Josie Godfrey
- ▶ **From Daily Life to Science: Gaps Between Everyday Life and Gene Therapy in Rare Diseases** – moderated by Yasemin Memişoğlu
- ▶ **Delivering equitable access for ultra-rare diseases across Europe** – moderated by Elena Nicod
- ▶ **From Invisible Symptoms to Measurable Outcomes: Overcoming Barriers in Collecting Patient-Reported Outcomes (PROMs) in Ultra-Rare Neurotransmitter Disorders** – moderated by Ana Pataki
- ▶ **Talking With Children About Rare and Hereditary Diseases: Practice and Evidence-Based Guidance for Families and Professionals** – moderated by Johanna Rintahaka
- ▶ **Basic funding of designated centers of expertise for Rare Diseases** – moderated by Claas Röhl
- ▶ **Preparing a Fit-for-Rare JCA to Power Competitiveness and Accelerate Access for the Rare Disease Patient Community** – moderated by Sophie Turner

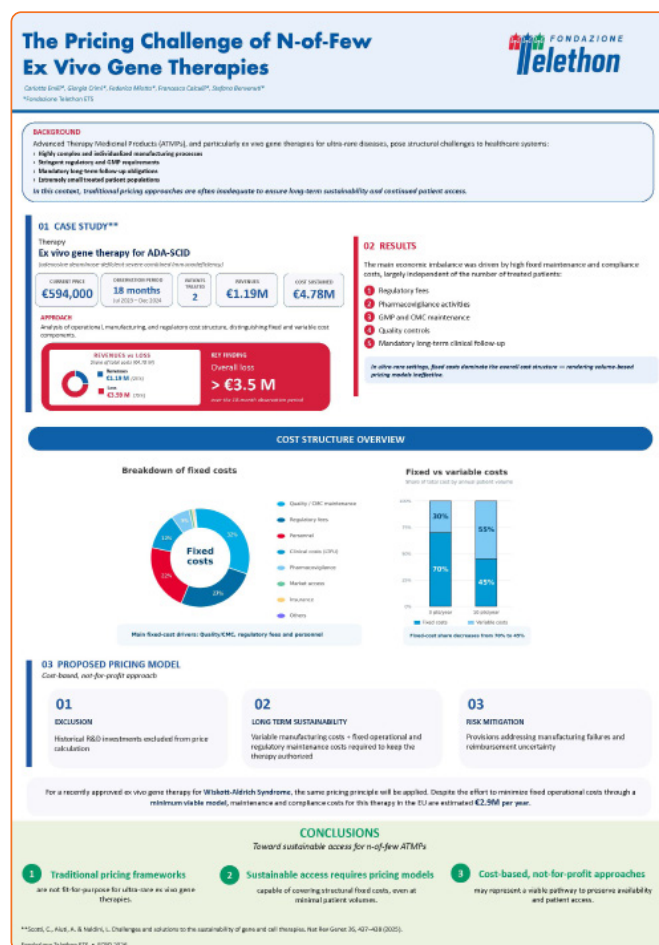


Over 200 poster abstracts were accepted for ECRD 2026 and displayed in an interactive virtual showcase, with over 100 posters exhibited on-site. The top 10 highest-scoring poster authors were invited to present their poster in dedicated poster pitch sessions, during which the audience was invited to vote for the poster they found the most impactful. The two winning posters this year were presented by Romain Alderweireldt, from the 101 Genomes Foundation, and by Carlotta Emili, from Fondazione Telethon.



## 96 – GEMS-App: A Web-Based Dynamic Consent Platform Enabling Genome-Wide Epistasis Research in Marfan Syndrome

Romain Alderweireldt  
101 Genomes Foundation



## 208 – The pricing challenge of an n-of-few ex-vivo gene therapy

Carlotta Emili  
Fondazione Telethon

[Read EURORDIS' article about the winning poster pitches!](#)

If you would like to find out more about the 200 + posters on display during ECRD 2026, you can access all via the following page: <https://www.rare-diseases.eu/posters/>

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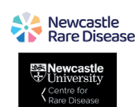
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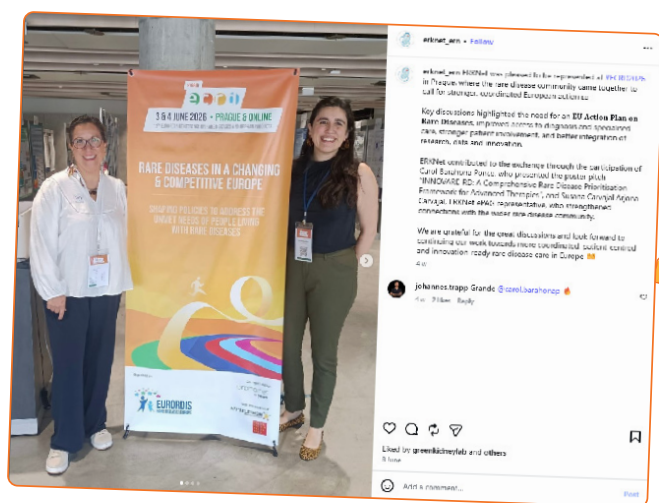
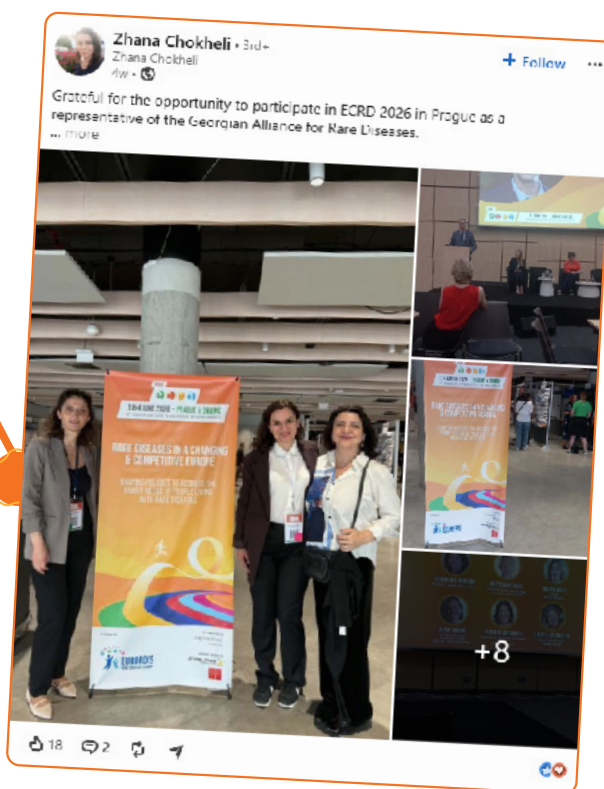
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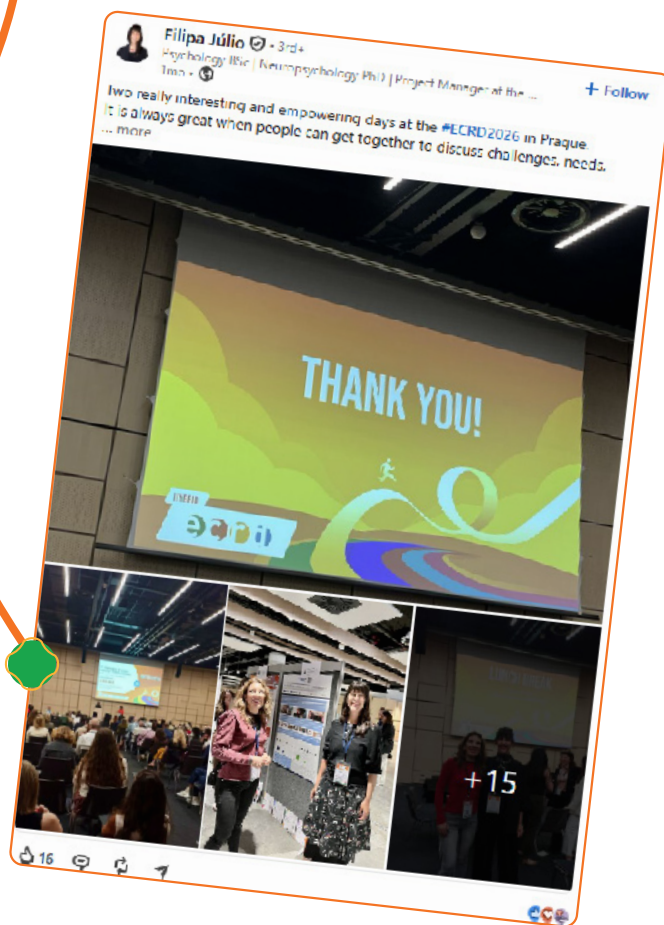
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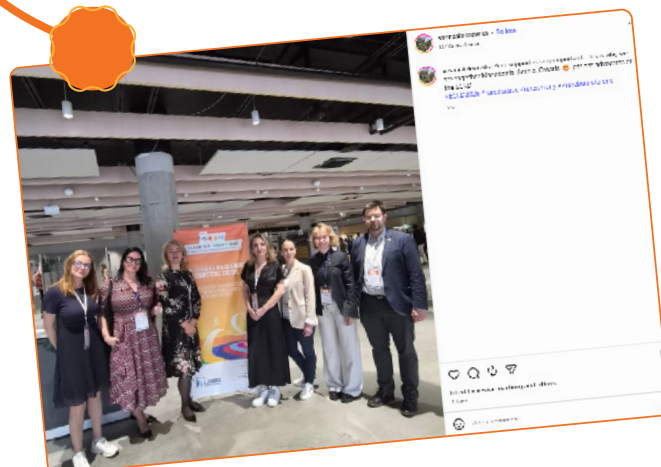
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