#RARE DISEASE DAY

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IMPACT OF RARE DISEASES: MORE THAN YOU CAN IMAGINE

EVENT REPORT

5 MARCH, 2025 13:30 – 15:30

JÓZSEF ANTALL 4Q1, EUROPEAN PARLIAMENT

Hosted by

MEP Stine Bosse, Renew Europe





MEP Adam Jarubas, EPP



Objectives of the meeting

To mark Rare Disease Day 2025 and following the launch of a new European Parliament Intergroup on Cancer and Rare Diseases, MEPs Stine Bosse (Renew, Denmark), Adam Jarubas (EPP, Poland) and EURORDIS-Rare Diseases Europe organised a special event entitled ***Impact of Rare Diseases: More than you can imagine***. This event aimed to recapitulate the issue of rare diseases to EU policymakers at the start of this new political mandate. In particular, it sought to **foster a deeper understanding of the challenges faced** by people living with rare diseases and their families, while presenting **concrete EU-level solutions** to address these issues effectively.

Taking place at the European Parliament in Brussels, this event was part of a broad range of activities **organised for the annual Rare Disease Day campaign**, a globally coordinated movement advocating for equal opportunities, healthcare access, and timely diagnosis and treatment for people living with rare diseases, regardless of where they live. This year's Rare Disease Day theme, "More than you can imagine," highlighted the **profound and far-reaching impact of rare diseases** on individuals, on their families, and on society as a whole, if no concrete measures are taken to address them.

Therefore, throughout the event, **thanks to the participation of key EU policymakers, experts and patient representatives**, we:

- Presented the **impact of living with a rare disease** on individuals and their families, while examining the broader effects of rare disease on healthcare systems, social structures, and society at large.
- Illustrated first-hand experiences with the **latest data and evidence**, offering a comprehensive understanding of the current realities faced by the rare disease community.
- Outlined **actionable policy solutions** at the European level, focusing on legislative and non-legislative measures that should be prioritised and adopted during this EU mandate.

Event Programme

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

Setting the scene - *13:30-14:00*

Introduction
Stine Bosse (Renew)
Introduction and welcome.

Vice Chair of the European Parliament's health committee (SANT) and member of the Renew group, Bosse welcomed attendees to the event, sharing her personal story about her daughter’s struggle with a rare viral brain infection, highlighting the lack of accessible treatment and research in Denmark at the time. This experience fuelled her passion for ensuring patients can access available treatments, especially for rare diseases. Bosse emphasised the need for collaboration among patients, advocates, clinicians, NGOs, and industry to address these challenges. She stressed the importance of European unity in providing healthcare solutions, even in difficult times, and called for increased funding and action to support vulnerable populations. Bosse also noted the recent U.S. policy changes on rare diseases and advocates for Europe to take a leading role in addressing these issues.

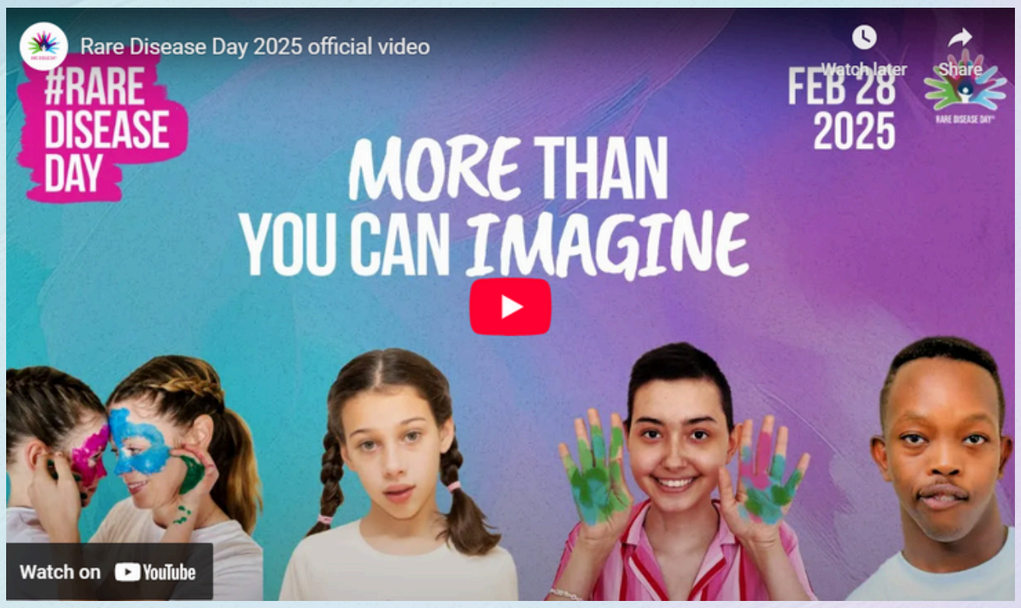
Key takeaways:

- Improving Access to Treatments for Rare Diseases: Ensuring patients across Europe can access available treatments, particularly for rare diseases, by addressing gaps in research, diagnosis, and healthcare infrastructure.
- Collaboration Across Stakeholders: Underlining the need for cooperation among patients, advocates, clinicians, NGOs, and industry to develop and deliver effective healthcare solutions.
- Increased Funding for Healthcare: Advocating for stronger financial support, including restoring and expanding health budgets in the EU, to address the needs of vulnerable populations and advance medical research and treatment accessibility.

Video of RDD

Introduced by Avril Daly, President, EURORDIS

EURORDIS 2025 Rare Disease Day video.



Watch the Rare Disease Day video here:

Setting the scene

Adéla Odrihocká, Rare Diseases Czech Republic

Reflection on key results from EURORDIS Rare Barometer surveys as seen through the lens of personal experience.

Adéla Odrihocká, representing the rare disease community, shared her personal experience of living with a rare disease, which took over a decade to diagnose. She highlighted the challenges faced by millions of Europeans, including delayed diagnoses, limited access to treatments, and inadequate social support.

Odrihocká emphasised that only 6% of rare diseases have causal treatments, and disparities in healthcare systems create significant barriers. She called for a coordinated European approach to improve diagnosis, strengthen research, and ensure affordable, accessible treatments. Odrihocká also stressed the need for person-centred, multidisciplinary care to address the emotional, financial, and social impacts on patients and their families.

She expressed hope in the progress made through European cooperation, such as cross-border expertise and treatment access, and urged for the implementation of the European Action Plan on Rare Diseases. She concluded by advocating for continued commitment to policies that ensure equal opportunities and support for those living with rare diseases.

Impact of Rare Diseases: facts and figures

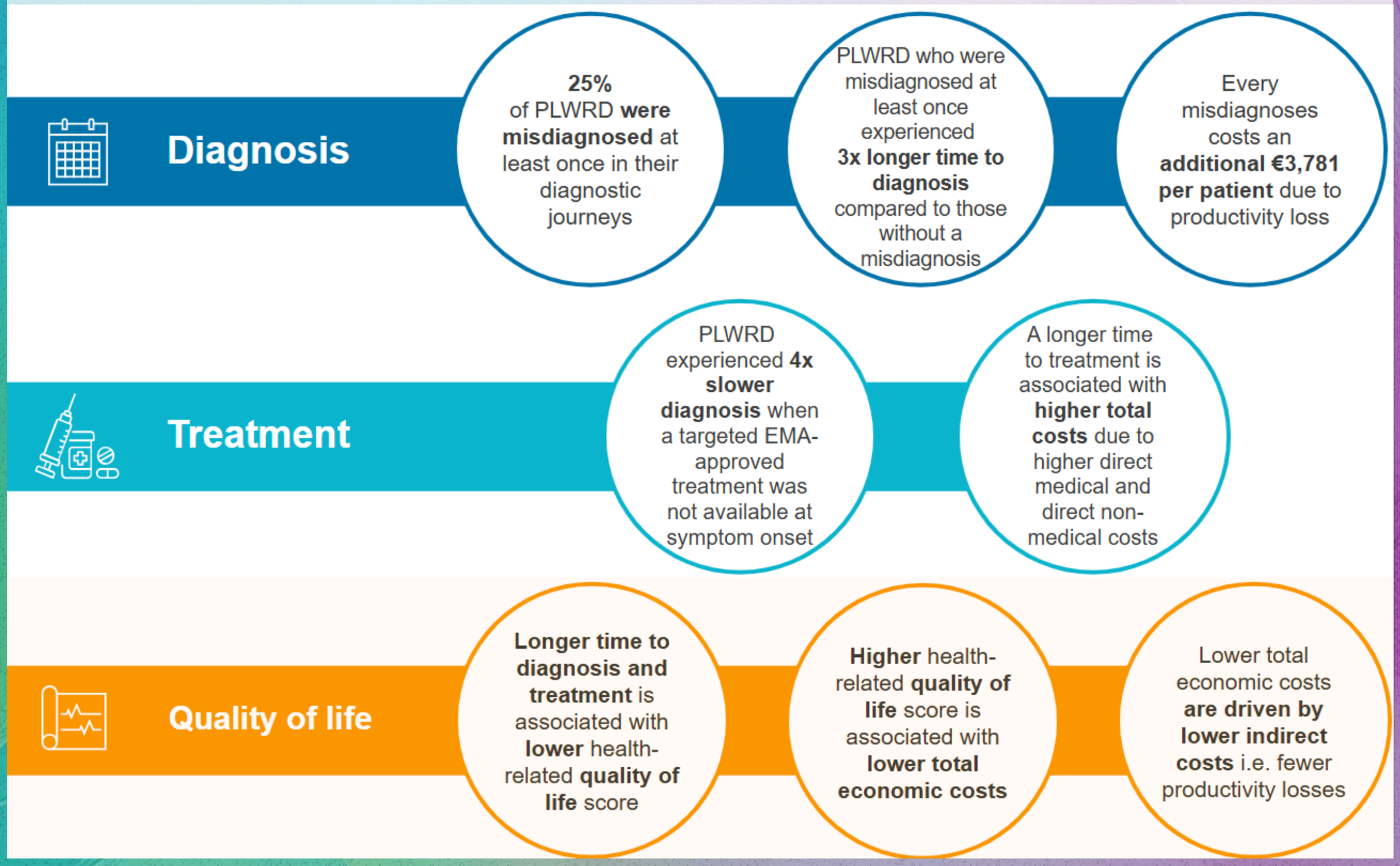
Tim Wilsdon, Vice President, Charles River Associates

Overview of key figures related to study on impact of RDs.

Tim Wilsdon presented key findings from a study on the economic cost of rare diseases across Europe, produced by Charles River Associates and Alexion, involving experts and patient surveys across nine countries and 43 diseases.

The study estimated the annual economic impact at around €250 billion, surpassing costs of cardiovascular diseases, oncology, and neurology. This cost includes direct medical expenses (74%, €184 billion), non-medical costs like caregiving (15%, €37 billion), and indirect costs such as lost productivity (11%, €27.5 billion). Rare disease patients face six times higher costs compared to those with non-rare diseases.

The study highlighted the diagnostic odyssey, with misdiagnoses leading to delayed treatment and higher costs. Faster diagnosis and access to treatments were shown to improve quality of life and reduce economic burdens. The findings underscore the need for holistic approaches, addressing healthcare, social, and labour market impacts, and prioritising faster diagnosis and treatment to benefit patients and reduce costs.



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Tripartite sessions - *14:00-15:10*

THE DIAGNOSIS OF RARE DISEASES TAKES

5 YEARS ON AVERAGE

Easier, faster and more accurate diagnosis of rare diseases

- *Olaf Riess, Coordinator, Solve-RD, and Centre for Rare Diseases, Tübingen*
- *Thomas Minten, KU Leuven*
- *MEP Vlad Voiculescu (Renew)*

The session on diagnosis, moderated by Enrique Terol, focused on the challenges and solutions for diagnosing rare diseases in Europe. Key speakers included Olaf Riess, Thomas Minten, and Vlad Voiculescu.

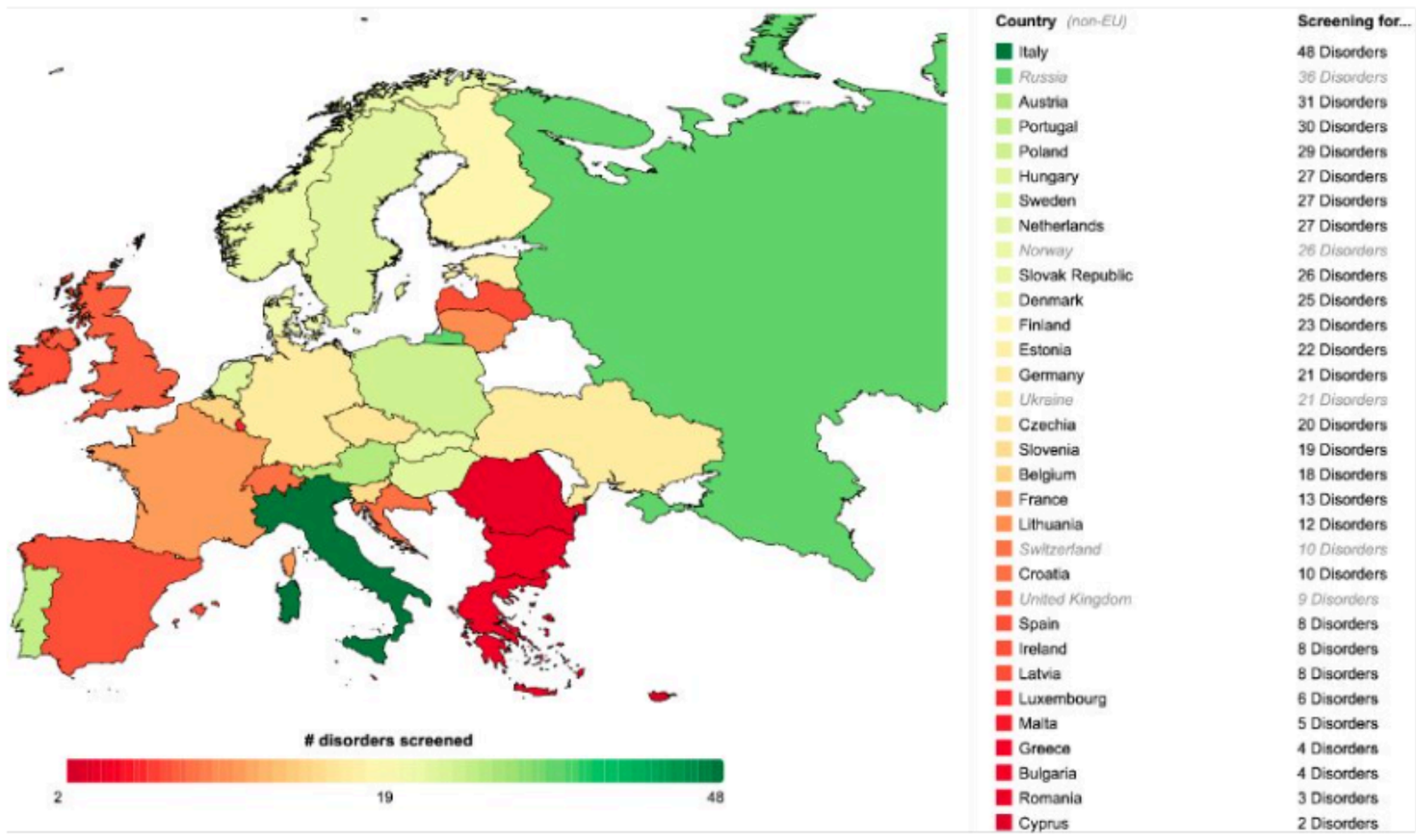
Riess highlighted the importance of advanced genomic technologies, such as whole-genome sequencing, to improve diagnosis rates, noting that 80% of rare diseases have genetic origins. He spoke of the need for better data sharing across Europe, overcoming data silos, and leveraging AI to integrate genetic, clinical, and imaging data for faster and more accurate diagnoses. He also stressed the role of European Reference Networks (ERNs) and the need for national-level coordination to ensure patient access to diagnostics across healthcare systems.

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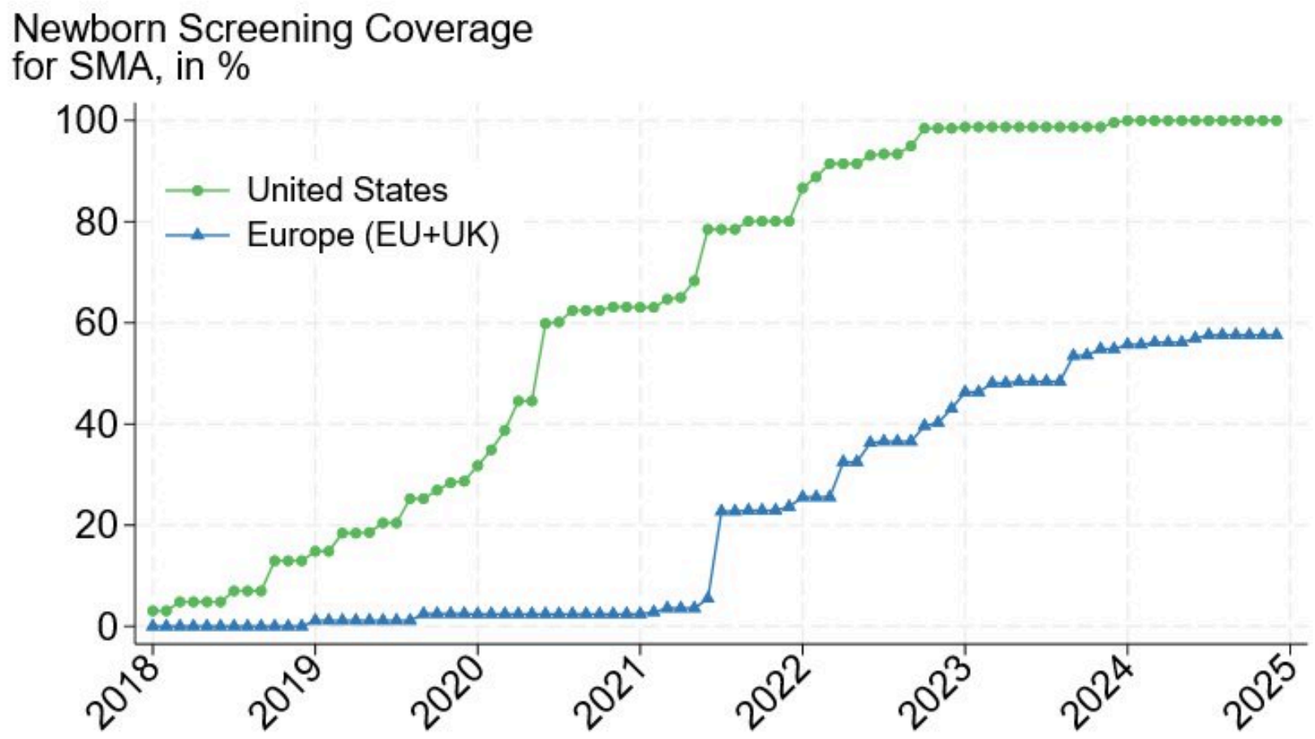
Thomas Minten discussed the disparities in newborn screening programmes across Europe, with the number of conditions screened ranging from 2 to 48 per country. He advocated for harmonised European guidelines to accelerate the adoption of life-saving screenings, using spinal muscular atrophy (SMA) as an example. Minten pointed out that delayed screening adoption in Europe has resulted in preventable disabilities for thousands of children, urging the EU to establish a centralised body to streamline newborn screening.

Newborn screening in Europe: a lottery



Sources:
(1) Charles River Associates (2024) A landscape assessment of newborn screening in Europe.
(2) Therrell et al. Current Status of Newborn Bloodspot Screening Worldwide 2024: A Comprehensive Review of Recent Activities (2020–2023). *International Journal of Neonatal Screening*. 2024; 10(2):38.

Example: slow SMA screening adoption



Comparison: ~1,298 more children diagnosed too late in EU+UK

Sources: US data from NewSTEPS APHL, EU data from SMA NBS Alliance and national websites. 1,298 is based on counterfactual calculations assuming Europe matched the US adoption rate with SMA prevalence 1 in 10,000.

Vlad Voiculescu, a Member of the European Parliament, underscored the importance of political will and cooperation to address diagnostic delays. He called for European-level strategies to improve diagnostic technologies, data sharing, and workforce training. Voiculescu highlighted the success of ERNs and cross-border healthcare initiatives, advocating for their expansion and increased funding. He also proposed practical steps, such as writing to national governments to push for unified newborn screening programmes.

Overall, the session emphasised the need for a coordinated European approach to reduce diagnostic delays, improve data accessibility, and invest in advanced technologies and workforce training to enhance the quality of life for rare disease patients.

Key Takeaways:

- Diagnostic Delays and Technology Gaps: Significant delays in diagnosing rare diseases, often taking years, due to limited access to advanced genomic technologies like whole genome sequencing and insufficient integration of diagnostic tools across healthcare systems.
- Data Sharing and Collaboration: The need to overcome data silos and improve data sharing across Europe, leveraging initiatives like the European Reference Networks (ERNs) and the European Health Data Space to enhance research and diagnosis.
- Harmonisation of Newborn Screening: Disparities in newborn screening programmes across EU countries, with calls for harmonised European guidelines to ensure timely and consistent screening for rare conditions.



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FEWER THAN

5%

OF KNOWN RARE DISEASES HAVE AN APPROVED PHARMACOTHERAPY

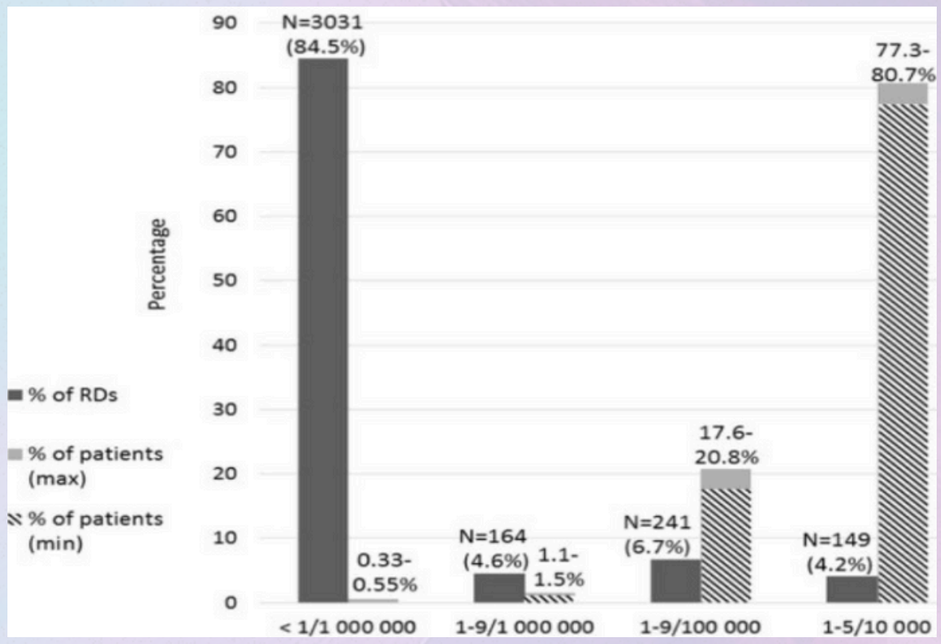
Development and Access to treatments for people living with rare diseases: EU cooperation and support needed

- *Sebastian Honoré, Co-Founder, Cure Lowe Foundation*
- *Stefano Benvenuti, IRDiRC member and Head of Public Affairs and Market Access, Fondazione Telethon*
- *MEP Stine Bosse (Renew)*

The session on access to treatment for rare diseases highlighted several critical issues and potential solutions.

Sebastian Honoré shared his personal journey as a father of a child with Lowe Syndrome, emphasising the economic barriers to developing treatments for ultra-rare diseases. He proposed solutions like widespread genetic testing, public-private partnerships, and a unified EU market to incentivise drug development.

Stefano Benvenuti from Fondazione Telethon discussed the challenges of maintaining treatments for ultra-rare diseases, citing the example of a gene therapy for ADA-SCID. He highlighted the unsustainable business model for ultra-rare diseases, where costs often exceed revenues, and called for regulatory reforms, centralised treatment centres, and improved cross-border patient mobility to ensure access.



Nguengang Wakap, S., Lambert, D.M., Olry, A. et al. Estimating cumulative point prevalence of rare diseases: analysis of the Orphanet database. Eur J Hum Genet 28, 165–173 (2020). https://doi.org/10.1038/s41431-019-0508-0

Stine Bosse MEP underlined the need for European collaboration to address rare diseases, given the low prevalence in individual countries. She advocated for de-risking investments through grants, equity support, and cross-border clinical trials. Bosse also stressed the importance of leveraging EU legislation, such as the Critical Medicines Act and Pharma Package, to create sustainable solutions and ensure patient access to treatments across borders.

The session underscored the need for innovative economic models, regulatory harmonisation, and cross-border healthcare coordination to improve access to treatments for rare disease patients in Europe.

Key takeaways:

- Limited Treatment Availability: Less than 5% of rare diseases have effective treatments, with access varying widely across EU member states, creating a "postcode lottery" for patients.
- Economic Barriers to Drug Development: High costs and low patient numbers make developing treatments for ultra-rare diseases economically unviable. Solutions proposed include transferable vouchers, public-private partnerships, and innovative funding mechanisms to de-risk investments.
- Need for EU-Wide Collaboration: Harmonising regulations, centralising patient registries, and improving cross-border healthcare access are essential to ensure equitable treatment access and sustainable business models for rare disease therapies.

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8 IN 10 PEOPLE WITH RARE DISEASES LIVE WITH DISABILITIES

Holistic care and support to address the impact of living with RDs

- *Adéla Odrihocká, Rare Diseases Czech Republic*
- *Karsten Vanden Wyngaert, Department of Nephrology, Ghent University Hospital and ERKNet Member*
- *MEP Tilly Metz (Greens)*

The session on holistic care for rare disease patients emphasised the need for comprehensive support beyond medical treatment, addressing physical, emotional, social, and financial challenges.

We heard again from Adéla Odrihocká, who shared her personal experience living with Ehlers-Danlos syndrome, highlighting the lack of state support, the overwhelming administrative burden of coordinating care, and the absence of multidisciplinary teams. She stressed the need for care coordinators to streamline access to health and social services.

Karsten Vanden Wyngaert stressed that holistic care must address all aspects of a patient's life, including mental health, employment, and social integration. He called for integrating social partners into healthcare systems and leveraging European Reference Networks (ERNs) to develop adaptable, feasible care pathways.

Tilly Metz MEP highlighted the intersection of rare diseases and disabilities, advocating for improved disability assessments, harmonised EU-wide disability recognition, and better access to employment and mental health support. She stressed the importance of systemic changes to empower patients and their families, ensuring they can participate fully in society.

The session underscored the urgent need for integrated, patient-centred approaches to improve the quality of life for rare disease patients across Europe.

Key takeaways:

- Comprehensive Support Needs: Rare disease patients face multifaceted challenges, including physical, emotional, social, and financial burdens, requiring holistic care that goes beyond medical treatment to include mental health support, employment accommodations, and social integration.
- Systemic Gaps and Administrative Burden: Patients often struggle with fragmented healthcare systems, lack of multidisciplinary care teams, and overwhelming administrative tasks to access services, highlighting the need for care coordinators and streamlined support systems.
- EU Policy and Integration: The session called for EU-wide policies to improve disability assessments, harmonise disability recognition, and integrate social partners into healthcare systems, ensuring patients receive comprehensive, coordinated care and can participate fully in society.



Concluding remarks - *15:10-15:30*

EESC Representative
Ágnes Cser, European Economic and Social Committee rapporteur
Institutional remarks linking to latest EESC Opinion on Rare Diseases.

More than you can imagine
Virginie Bros-Facer, CEO, EURORDIS-Rare Diseases Europe
Summary of cross-cutting issues and need for EU Action Plan, as well as continued financial support in the upcoming MFF.

Concluding remarks
MEP Stine Bosse (Renew)
Wrap-up and goodbye.

The closing session highlighted the importance of continued collaboration and momentum in addressing rare diseases.

Ágnes Cser called for 2026 to be designated as the "Year of Rare Disease Patients" and urged support for the European Action Plan to achieve faster, smarter, and more cost-effective solutions.

Virginie Bros-Facer thanked the MEPs for hosting the event and highlighted the patient community's readiness to contribute with evidence and data.

Stine Bosse concluded by affirming the commitment to listen and act, encouraging ongoing advocacy and cross-border cooperation to drive progress for rare disease patients and their families.

