

Pre-ECRD Workshop:

European Blueprint for Rare Diseases

Context and rationale of the Workshop

Rare diseases affect more than 30 million people across Europe, yet significant disparities persist in diagnosis, care, research, and access to treatments between and within countries. While considerable progress has been achieved over recent decades, people living with a rare disease and their families continue to face substantial challenges that impact their health outcomes, quality of life, and social inclusion.

To address these ongoing gaps, **EURORDIS – Rare Diseases Europe has convened a multi-stakeholder Core Group (CG)** and, in collaboration with Rare Diseases International (RDI), **a European Regional Task Force (RTF)** to develop a **European Blueprint for Rare Diseases**. The Blueprint seeks to bring together existing evidence, policy recommendations, and stakeholder expertise into a **coherent and actionable framework capable of guiding future rare disease policy development at both European and global levels**. It is intended to serve as the foundation for a **future EU Action Plan on Rare Diseases** while also informing the development of a **Global Rare Disease Action Plan** for the WHO European Region.

As part of this process, a **dedicated Pre-Conference Workshop** was held on 2 June 2026 in Prague, ahead of the European Conference on Rare Diseases and Orphan Products (ECRD 2026). With over 150 registered participants, the workshop represented a significant milestone in the development of the European Blueprint for Rare Diseases. It provided a **dedicated forum for members of the Core Group, the Regional Task Force, and other key stakeholders to examine and discuss the proposed policy actions and measures underpinning the Blueprint**.

The workshop was designed to gather final feedback on the proposed actions, identify priorities, explore areas of consensus and divergence, and strengthen stakeholder ownership of the Blueprint ahead of its next phase of development. Through a combination of plenary sessions and thematic breakout discussions, participants examined six key areas:

1. Integrated and person-centred care and inclusion in society;
2. Innovative and needs-led research;
3. Access to treatments and medical devices; earlier and more accurate diagnosis;
4. Enabling faster, more accurate diagnosis
5. Access to specialised healthcare;
6. Optimisation of the rare disease data ecosystem

In their breakout sessions, stakeholders from across Europe analysed and discussed the actions proposed. Through the support of a facilitator, they were asked to perform a prioritisation exercise. Some elements were presented to all participants as possible criteria guiding their judgment:

- the **extent** to which the action addresses major unmet needs or inequities for people living with a rare condition;
- the **severity** and scale of the problem it aims to address;
- the **expected** impact of the action if effectively implemented;
- whether the action could **enable progress** in other areas.

The discussions focused on identifying **priority actions for both a future EU Action Plan and a Global Action Plan on Rare Diseases**, while also capturing the criteria and rationale used by participants in their prioritisation process. Most groups proposed amendments to the actions, including merging, streamlining or revising the existing text. All reflections and contributions were recorded by the note-takers and will inform the finalisation of the European Blueprint for Rare Diseases, alongside feedback received from the Core Group and the Regional Task Force.

In this report

In the following sections, each of the six discussion tables is presented, highlighting the actions identified as priorities at both EU and global levels, as well as the key discussion points and insights that emerged during the exchanges.

For reference, the full list of policy actions presented to participants for prioritisation is available via the hyperlink provided at the beginning of each section.

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1. Integrated and person-centred care and social inclusion

The Group discussed this set of proposed action for the *Integrated Care and Social Inclusion* pillar of the European Blueprint for Rare Diseases.

Actions selected as EU priorities:

1. Develop EU guidance and implementation tools for Member States for multidisciplinary, person-centred rare disease integrated care pathways (including social care)

7. Integrate social services into care for patients with rare diseases and recognise those living with rare diseases as a disadvantaged group, ensuring that they receive appropriate allowances and benefits, much in the same way as people with disabilities (with link to Action 8 and 9)

10. Develop practical guidance and monitoring instruments for Member States to enable them to embed reasonable adjustments in mainstream education and employment policies

Actions selected as Global priorities:

1. Develop EU guidance and implementation tools for Member States for multidisciplinary, person-centred rare disease integrated care pathways (including social care)

7. Integrate social services into care for patients with rare diseases and recognise those living with rare diseases as a disadvantaged group, ensuring that they receive appropriate allowances and benefits, much in the same way as people with disabilities

10. Develop practical guidance and monitoring instruments for Member States to enable them to embed reasonable adjustments in mainstream education and employment policies

Discussion

The discussion centred on the need to move beyond fragmented health and social services towards **a genuinely integrated, person-centred model of care** that addresses the full range of needs experienced by people living with rare diseases and their families. The group agreed that outstanding priorities in the areas are the same for both the EU and Global Rare Diseases Action Plans.

While participants discussed a broad range of actions, there was strong convergence around the importance of **integrated care pathways** (Action 1), **psychosocial and practical support** throughout the patient journey (Action 7), and stronger access to **social protection and disability-related services** (Actions 8, 9 and 10). Participants repeatedly

emphasised that rare diseases cannot be addressed solely through healthcare systems and that effective support requires coordinated action across health, social care, education, employment and community services. Several contributors highlighted the need for stronger links between ministries responsible for health and social affairs, noting that many existing systems (Italy was raised as an example) continue to treat rare diseases and disability as separate policy domains, creating barriers to access and continuity of support.

Participants stressed that integrated care pathways should be composed of multidisciplinary teams supporting individuals across their entire disease journey. Particular attention was given to the long diagnostic odyssey experienced by many patients and families, with participants arguing that psychosocial and practical support should be available before **diagnosis**, during diagnosis and throughout long-term care. Mental health support, counselling, treatment education and coordination assistance were consistently identified as essential components of care, with several participants emphasising that support should extend to family members and informal carers as well as patients themselves. This strong support resulted in **psychosocial** and practical support services (Action 7) emerging as the highest-ranked priority during voting.

The discussion also highlighted the need for **better coordination and continuity of care** across different stages of life and between different services, including structured transition processes from paediatric to adult services. The **transition period** was repeatedly cited as a point of vulnerability where patients often experience disruptions in care and support. In this context, participants viewed Actions 1, 5 and 7 as closely interconnected and proposed grouping them together as part of a broader approach to integrated, lifelong care. The importance of **case management** was also recognised, with participants supporting both the development of EU-level guidance on case management and investment in training programmes to professionalise and strengthen this function across Member States.

Another major theme concerned **social inclusion**, disability recognition and access to social protection. Participants argued that many people living with rare diseases face significant functional limitations and socioeconomic disadvantages that are not adequately recognised by existing **disability assessment systems**. There was broad support for developing EU guidance on disability assessment procedures (Action 8), particularly to ensure that rare disease-related impairments, including those associated with undiagnosed conditions, are recognised in a consistent and timely manner. Participants linked this closely to the need to **integrate social services into care pathways** (Action 9) and to guarantee access to appropriate social protection measures, social workers, allowances and support services tailored to individual needs (Action 10). These actions were widely viewed as inseparable components of a comprehensive framework for social inclusion and independent living and were therefore grouped together during the prioritisation exercise.

Another strong message emerging from the discussion was the **value of lived experience** in shaping policy and service design. Participants stressed that people living with rare diseases, families and carers should be involved in governance, implementation and

evaluation processes. Suggestions included creating formal structures through which people with lived experience could work alongside clinicians and policymakers to influence decisions at national and European levels. This emphasis reflected a broader recognition that effective care must be informed by the realities of daily life with a rare disease and not solely by institutional or clinical perspectives.

Finally, participants highlighted the need for **stronger evidence on the socioeconomic burden of rare diseases**. Action 13 received considerable support, with participants arguing that better data on social, economic and quality-of-life impacts would strengthen policy development, resource allocation and advocacy efforts. The discussion recognised that many of the challenges faced by people living with rare diseases remain insufficiently documented and that robust evidence is needed to demonstrate the full costs associated with fragmented care, unmet social needs and barriers to participation in society.



Overall, the workshop articulated a vision of integrated care and social inclusion built around four interconnected priorities, equally important for countries within the European Union and outside: **multidisciplinary and person-centred care pathways, comprehensive psychosocial support, equitable access to social protection and disability recognition, the integration of rare disease-specific accommodations into education and employment policies, and meaningful involvement of people with lived experience.**

List of people registered for the discussion:

Name	Surname	Affiliation
Jessie	Dubief	EURORDIS (Rare Diseases Europe) -FACILITATOR
Adéla	Odrihocká	ČAVO (Czech Association for Rare Diseases) – Notetaker
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Dorica	Dan	Romanian National Alliance for Rare Diseases (RONARD) / EURORDIS
Iolo	Eilian	Health Service Executive (HSE) Ireland - Rare Disease Office
Charissa	Frank	ERN ReCONNET (ePAG Representative for Ehlers-Danlos Syndromes)
Kirsten	Johnson	Fragile X Society (UK) - EURORDIS (Rare Diseases Europe)
Filipa	Júlio	European Huntington Association (EHA)

Name	Surname	Affiliation
Maria Luisa	Scattoni	Istituto Superiore di Sanità (ISS) - National Center for Rare Diseases
Aleksandra	Milutinovic	National Organisation for Rare Diseases Serbia (NORBS)
Ida	Mirković Knaus	Croatian Alliance for Rare Diseases (HSRB)
Pauline	Nauroy	Fondation Maladies Rares (French Foundation for Rare Diseases)
Anders	Olauson	Ågrenska (National Competence Centre for Rare Diseases, Sweden)
Greet	Op de Beeck	Radiorg (Belgian Rare Diseases Federation)
Christel	Paris-Bicking	Mental ID (Health Innovation & Support Platforms)
Ana	Pataki	Lil' Brave One - Hrabriša (Neurotransmitter Disorders Advocacy) / EURORDIS
Annalisa	Scopinaro	UNIAMO (Federazione Italiana Malattie Rare)
Suja	Somanadhan	University College Dublin (UCD) - Rare Disease Nursing Research
Kateřina	Strnadová	Rett Community Czechia (Rett-CZ)
Alessandra	Tolaccia	OIFE (Osteogenesis Imperfecta Federation Europe)
Lex	van der Heijden	CMTC-OVM (Vascular Malformations Patient Organisation)
Danica	Vučić	National Organisation for Rare Diseases Serbia (NORBS Help Line)
Hind	El Amiri	European Brain Council (EBC) / Rare Disease Research Liaison
Stefan	Zivkovic	National Organisation for Rare Diseases Serbia (NORBS) / PH Serbia

2. Research and innovation

The Group discussed this set of proposed action for the [Research and Innovation](#) pillar of the European Blueprint for Rare Diseases.

Actions selected as EU priorities:

4. Strengthen structured and meaningful patient involvement in EU-funded rare disease research (including priority setting, governance of projects, protocol design, endpoints, dissemination) to better align R&D with patient needs and incentivise the use of similar patient involvement guidelines at national level. This should include the development of clear guidance for patient involvement and the provision of adequate resources for patient organisations to build capacities and provide input.

7. Strengthen interoperable EU research and clinical infrastructures (registries, biobanks, trial networks) with common standards and sustainable governance.

2. Establish sustained EU co-investment mechanisms for rare disease research, aligned with patients' needs, across the full pathway, leveraging public–private models, existing rare disease research infrastructures, and long-term partnerships + **11. Promote innovative trial designs and regulatory science** for small populations (adaptive/platform trials, external controls), with early dialogue and learning cycles

Actions selected as Global priorities:

12. Support the deployment of digital platforms for remote participation in studies and follow-up, reducing burden for patients and carers

Discussion

The discussion was dominated by a shared recognition that rare disease research requires a more **coordinated, sustainable and patient-centred ecosystem**. While participants broadly supported the idea of a European Rare Disease Research Strategy (Action 1), the prevailing view was that **developing an overarching strategy is not an objective in itself** unless it is translated into concrete mechanisms for funding, infrastructure, evidence generation and implementation. A clear trend emerged towards prioritising actions that support the entire research pathway—from discovery to patient benefit—rather than focusing on isolated research projects.

One of the strongest themes was the need for **long-term, sustainable investment**. Participants repeatedly emphasised that funding should not only support research projects but also preserve and strengthen the infrastructures that make research possible, including registries, biobanks, datasets, trial networks and expert collaborations. There was concern that **valuable assets are frequently lost when project funding ends**, highlighting the need to move away from short-term project cycles and towards a more durable research ecosystem. In this context, investment (Action 2) was generally perceived as a **necessary enabling condition** for supporting infrastructure, trial readiness, regulatory science, public–private collaboration and implementation. While the group did not identify support for Public–Private Partnerships (Action 9) as a standalone priority, it recognised this as a crucial component of a broader **ecosystem of sustainable co-investment**.

The themes of investment and clinical trial design were closely linked throughout the discussion. A sustained EU co-investment mechanism was identified as complementary to innovative trial designs for small populations. The discussion highlighted the need to rethink research and development models for rare and ultra-rare diseases. Innovative trial designs, adaptive methodologies, platform trials and regulatory approaches tailored to small populations were widely viewed as critical to accelerating progress. Many promising innovations fail to progress because conventional commercial incentives are insufficient, leading to calls for dedicated pathways for ultra-rare conditions, N-of-1 therapies, repurposing and other commercially fragile areas (Action 15).

A further recurring theme, which generated strong consensus among participants, was the **recognition that data and infrastructure are foundational enablers of progress.** Interoperable registries, biobanks, trial networks, common standards and sustainable governance were repeatedly described as prerequisites for achieving many other ambitions, including innovative trial designs, artificial intelligence, regulatory innovation and international collaboration. Participants consistently argued that fragmented data systems remain a major obstacle and that strengthening European and cross-border infrastructures should be viewed as a **strategic priority**, given the barriers fragmentation creates for both academic research and commercial viability. Although support for Centres and Networks (Action 8) was not ultimately prioritised, many participants argued that it should be considered part of the implementation of Action 7 and called for the two actions to be merged. Similarly, sustaining the funding and operational capacity of European Reference Networks (ERNs) (Action 10) was seen as an important step. However, participants stressed the need to clarify how ERNs contribute to research infrastructure, trial readiness and evidence generation.

Patient participation emerged as another central priority and was consistently discussed as a **fundamental shift rather than a procedural requirement.** Participants stressed that patients should be embedded throughout the research process, from priority-setting and governance to trial design, outcome measures, evidence generation, dissemination and implementation. Closely linked to this was strong support for **patient-generated evidence**, including patient-reported outcomes, experience measures and quality-of-life data. These were viewed as essential for understanding disease burden, identifying unmet needs and ensuring that research reflects real-world priorities. The corresponding Action 12 was selected as a global priority.

Participants also emphasised the importance of international collaboration, recognising that the small and geographically dispersed nature of rare disease populations requires research efforts that extend beyond national borders. Multi-country studies, shared protocols and interoperable infrastructures were viewed as essential for generating robust evidence and reducing inequalities in access to research opportunities.



Overall, the workshop highlighted a vision of rare disease research built on four interconnected pillars: **sustained investment, meaningful patient partnership, interoperable research infrastructures and innovative development pathways for small populations**. The key recommendation was to prioritise the enabling conditions that strengthen the entire research ecosystem, ensuring that scientific advances are translated more effectively into therapies, evidence and tangible benefits for people living with rare diseases.

List of people registered for the discussion:

Name	Surname	Affiliation
Roseline	Favresse	EURORDIS (Rare Diseases Europe) - FACILITATOR
Julien	Poulain	EURORDIS (Rare Diseases Europe) - Notetaker
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Anne-Mette	Bredahl	VASCERN / National Centre for Rare Disorders Norway
Giorgia	Buoncuore	Istituto Superiore di Sanità (ISS) - National Center for Rare Diseases
June	Buvarp	Independent Patient/Clinical Consultant (Rare Diseases)
Seán	Byrne	European Federation of Pharmaceutical Industries and Associations (EFPIA)
Donatella	Capone	NaNa Onlus (Associazione Nazionale Neurofibromatosi)
Aldona	Chmielewska	Agora Research
Emilia	Debska	SMA Europe
Claire	Duggan	Retina International
Shota	Dzemaili	University College Dublin (UCD)
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Ellen	Koekoekx	Cure Angelman Belgium
Malgorzata	Kosla	Rare Disease Patient Advocate / Consultant

Name	Surname	Affiliation
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Vitaliy	Matyushenko	CSMA Ukraine (Children with Spinal Muscular Atrophy)
Lubomír	Mazouch	Rare Disease Patient Representative / Advocate (Czechia)
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Ana	Moreira	International VATPase Alliance
Amanda	Rohrig	BridgeBio Pharma
Marion	Rottenberg	AOP Health
Stephanie	Sangalang	University College Dublin (UCD)
Rachel	Smith	Parexel (Rare Disease Clinical Trials)
Dorit	Wahl-Brink	Sjældne Diagnoser (Rare Diseases Denmark)
Stephen	Waldek	University of Sunderland (Lysosomal Storage Disorders Specialist)

3. Access to treatments and medical devices

The Group discussed this set of proposed action for the [Access to treatments and Medical devices](#) pillar of the European Blueprint for Rare Diseases.

Actions selected as EU priorities:

5. Support the sharing of best practices between Member States on early access principles for rare disease therapies (managed access, conditional reimbursement, data collection) and to ensure early access schemes include patient-informed criteria and transparent communication on eligibility and timelines.

14. Pilot HTA approaches adapted to rare diseases, including acceptance of alternative evidence, surrogate endpoints and iterative reassessment.

6. Strengthen cross-border access to specialised treatments and devices by improving pathways and coordination between centres and payers, and by streamlining prior authorisation, reimbursement, and referral processes at EU and national levels.

Actions selected as Global priorities:

13. Establish a permanent, structured early dialogue mechanism spanning the full development-to-access pipeline, bringing together developers, regulators, HTA bodies, payers, and patients to align evidence requirements and support patient partnership in access and reimbursement pathways.

14. Pilot HTA approaches adapted to rare diseases, including acceptance of alternative evidence, surrogate endpoints and iterative reassessment

9. Ensure continuity of access to treatments across life stages, including structured transition pathways from paediatric to adult care and harmonised treatment eligibility criteria across Member States.

Discussion

The discussion on access to treatments and medical devices was dominated by concerns about **inequities in access**, both between and within countries. A clear trend emerged towards viewing the **treatment pathway as a continuum** extending from regulatory approval and evidence generation to reimbursement, cross-border access, and long-term patient outcomes. This was deemed particularly relevant at global level (Action 9), with participants highlighting how it is a cross-cutting problem across states and diseases.

Several speakers argued that policy discussions often **focus too heavily on authorisation while neglecting the barriers that arise after approval**, particularly in relation to pricing, reimbursement, and national implementation. Indeed, a major theme was the need to

modernise regulatory, HTA, and reimbursement frameworks to better reflect the realities of rare diseases. The proposal of piloting new HTA approaches for rare diseases (Action 14) gained particular traction and chosen as a priority both at EU and Global level. Participants highlighted the limitations of conventional evidence requirements for small populations and called for greater flexibility across regulatory authorities, HTA bodies, and payers. The discussion consistently linked evidence generation, real-world evidence, conditional reimbursement, and adapted HTA methodologies as part of a broader effort to create more appropriate access pathways for rare and ultra-rare conditions. **Early access schemes** were particularly valued because of their potential to deliver immediate benefits to patients while supporting evidence generation and reimbursement discussions. **Patient participation** was consistently described as essential within these processes, particularly in HTA and reimbursement discussions.

Cross-border collaboration emerged as another dominant trend, the action proposing sharing of best practices and early access principles across Member States (5) received strong support as a mechanism to reduce disparities and promote harmonisation. Participants repeatedly referred to disparities in access between Member States and recognised that many countries, particularly smaller ones, lack sufficient patient populations, expertise, or infrastructure to address rare diseases independently. Facilitate **voluntary joint procurement and negotiations** was a topic of discussion both for the EU and global level, some participants emphasised its importance for smaller EU member states and ultra-rare, asking to link this proposal to action 6 (cross-border healthcare) and action 18 (EU Rare Disease Innovation and Access Fund under the next MFF).

The importance of multi-stakeholder dialogue was another recurring message. Participants strongly supported the establishment of **structured mechanisms** bringing together regulators, HTA bodies, payers, developers, clinicians, and patients across the entire development-to-access pathway. **Early dialogue** was viewed as a practical way to reduce bottlenecks, align evidence requirements, improve predictability, and address inequities between healthcare systems with different levels of maturity. The importance of establishing permanent and structured forms of early dialogue mechanisms at global level emerged clearly by the interventions (Action 13). Participants linked this to the necessity of involving patients

A further trend was the growing recognition that access challenges cannot be separated from broader questions of **sustainability and value**. Participants discussed the need to better demonstrate the **long-term societal and healthcare benefits** of treatment, arguing that therapies should be viewed as investments rather than costs. There was particular interest in generating evidence after approval, **improving post-marketing data collection**, and using **real-world evidence to support reimbursement decisions** and managed entry agreements (Action 15). The proposal for stronger incentives for evidence generation was described as an enabler for a more efficient HTA exercise, addressing the need for access,

improving dialogue between regulators and companies, but also providing more data for evidence-based reimbursement decisions.



Overall, the workshop called for treating **access as a shared responsibility across the entire ecosystem**. Rather than isolated policy interventions, it advocated integrated frameworks linking regulatory innovation, evidence generation, HTA, reimbursement, cross-border cooperation, patient involvement and sustainable funding. The overarching message was that reducing inequities requires coordinated action across the entire treatment pathway, supported by stronger collaboration between countries and stakeholders.

List of people registered for the discussion:

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Rene	Břečťan	ČAVO (Czech Association for Rare Diseases)
Daniel	de Vicente	FEDER (Spanish Federation for Rare Diseases)
Dante	Di Iulio	Chiesi Farmaceutici
Nancy	Di Salvo	GBS - CIDP Foundation International
Mariette	Driessens	VSOP (Dutch Patient Alliance for Rare and Genetic Diseases)
Tomasz	Grybek	Fundacja Badań i Rozwoju (B&R Foundation)
Florian	Innig	ACHSE (German Alliance for Chronic Rare Diseases)
Stan	Jackson	Astellas Pharma
Anneliene	Jonker	University of Twente (Technical Medical Centre)
Eka	Kandelaki	Tbilisi State Medical University (TSMU)
Aleksandra	Leijenhorst	Independent Rare Disease Consultant / Advocate

Name	Surname	Affiliation
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Jean	Saslawsky	EURORDIS (Rare Diseases Europe)
Roudie	Shafie	Ultragenyx Pharmaceutical
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Chris	Sotirelis	Thalassaemia International Federation (TIF) / Patient Advocate
Leo	Strican	Acadia Pharmaceuticals
Harry	Thurston-Smith	Jazz Pharmaceuticals
Dorota	Zgodka	Health Technology Assessment international (HTAi)

4. Enabling earlier, faster, and more accurate diagnosis

The Group discussed this set of proposed action for the *Diagnosis* pillar of the European Blueprint for Rare Diseases.

Actions selected as EU priorities:

- 8. Establish an EU-level newborn screening (NBS) advisory or multistakeholder group to provide non-binding recommendations on screening panels, quality standards, and ethical safeguards.
- 15. Establish structured pathways for undiagnosed patients, including referral to specialised centres and periodic reanalysis of genomic data.
- 18. Fund comparative research on diagnostic performance across Member States to identify gaps and guide EU action and develop consensus indicators (e.g. via OMC) to monitor diagnostics, care, research, and wellbeing.

Actions selected as Global priorities:

- 5. Fund a programme to strengthen professional education on rare diseases, including familiarising with the national network of CoEs in primary care and emergency settings, using case-based training and e-learning, dissemination of targeted campaigns and digital tools (e.g. the app developed by ERNs and Orphanet)
- 16. Support patient registries and data sharing to improve matchmaking and knowledge generation for undiagnosed conditions.

Discussion

The discussion showed strong consensus that earlier and more accurate diagnosis remains the cornerstone of rare disease policy, but participants repeatedly stressed that **diagnosis should not be treated as an end in itself**. A clear trend emerged towards viewing diagnosis as part of a **continuous patient journey** that includes care coordination, social support, long-term follow-up, and quality of life outcomes. Across interventions, there was broad agreement that the real measure of success is not simply obtaining a diagnosis, but ensuring that diagnosis leads to meaningful improvements for patients and families. The need for implementing a structured diagnosis pathway (Action 1) was judged as a priority for the Global level, while the need to embed case management and coordinate referral in such pathways (Action 2) was ultimately seen as a highly practical recommendation with great benefit and implications for the quality of lives of patients and their caregivers (especially for the EU).

Another recurring theme was the need for greater integration and coordination. Participants highlighted **growing fragmentation** between rare disease plans, genomics strategies, newborn screening initiatives, and other policy frameworks, calling instead for **an overarching approach that aligns existing efforts and avoids duplication**. Several speakers emphasised that structured diagnostic pathways, clear referral mechanisms, and stronger links between primary care, specialist centres, and European Reference Networks are essential to reducing delays and inequalities across countries. Great attention was reserved also to strengthening genomic counselling (Action 4, 9, 10, 11), and the relative measures needed for increasing the number of centres and professional offering such service.

Establishing an **EU-level newborn screening (NBS) advisory or multistakeholder group** was voted as an EU-level priority, reflecting a broad consensus that greater coordination is needed to address the significant variation in NBS programmes across Member States. Such a framework could support more equitable access to early diagnosis by facilitating the exchange of best practices, scaling up successful national pilot projects, and providing guidance on the evaluation and adoption of new technologies. Several contributors stressed that the body should ensure **meaningful patient involvement and support the development of harmonised approaches** to follow-up care, recognising that screening alone does not improve outcomes unless it is linked to appropriate treatment, care pathways, and long-term support. At the same time, participants cautioned against an **overly narrow focus on newborn screening**, noting that many rare diseases are diagnosed later in life and that strategies for earlier diagnosis must extend beyond the neonatal period.

The discussion also highlighted the need to increase attention to **undiagnosed patients**. Rather than viewing them as a separate population, participants described undiagnosed individuals as being at a different stage of the same diagnostic continuum. The need to establish structured pathways for undiagnosed patients (Action 15) received attention, especially at EU level. A few participants highlighted the resistance in Member States to define and address the needs of undiagnosed patients, making them even less visible in the health systems. This led to calls for structured pathways for unresolved cases, periodic reanalysis of genomic data, stronger cross-border collaboration, and improved visibility of undiagnosed conditions.

Education and awareness emerged as one of the strongest priorities, particularly at the global level (Action 5). Participants consistently noted that lack of awareness among healthcare professionals remains a major barrier to timely diagnosis and contributes to the isolation experienced by many people living with rare diseases. Supporting the creation of patient registries (Action 16) was extensively prioritised for the global level, particularly for the great implications it might have for knowledge generation in a field with such scattered population as rare diseases.

Finally, there was strong **support for investments in data infrastructure, registries, data sharing, and emerging technologies such as AI and multi-omics** (Action 18). However,

speakers stressed that these tools should be implemented within robust governance frameworks and used as enablers of broader systemic change.



The overall recommendation was **to prioritise strategic interventions that create and enable further advancements**, laying down not only responses to present challenges but the basis for **tomorrow's necessities and emerging technologies**.

List of people registered for the discussion:

Name	Surname	Affiliation
Gulcin	Gumus	EURORDIS (Rare Diseases Europe) - FACILITATOR
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Susana	Susan	Rare Disease Patient Representative / Consultant
Zhana	Chokheli	Tbilisi State Medical University / Rare Disease Advocate
Marian	Ciopec	EPODIN (European Patient Organisation for Dysimmune & Inflammatory Neuropathies)
Morgane	Cuisenier	Novo Nordisk (Rare Endocrine / Rare Diseases Division)
Andrew	Dempsey	UCB Pharma (Rare Disease Division)
Amalia E.	Gentile	Istituto Superiore di Sanità (ISS) - National Center for Rare Diseases
Sara	Elgott	Opal Medica
Holm	Graessner	Centre for Rare Diseases Tübingen / Coordinator of ERN-RND
Urh	Groselj	University Medical Centre Ljubljana (Pediatric Endocrinologist / Rare Diseases)
Anne	Hugon	AP-HP (Assistance Publique - Hôpitaux de Paris / Rare Diseases)
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Name	Surname	Affiliation
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Gergely	Meszaros	ERN-LUNG (European Reference Network for Rare Respiratory Diseases)
Alanna	Miller	Rare Diseases International (RDI)
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Hafdís	Ragnarsdóttir	Einstök Börn (Rare Diseases Iceland)
Maud	Maud	CSMA Ukraine (Children with Spinal Muscular Atrophy Advocacy)
Eva-Maria	Strömsholm	Harvinaiset (Rare Diseases Finland Support Network)
Justyna	Walczuk	MEK2 Research (Rare Disease Clinical & Genetic Research)

5. Access to specialised and highly specialised healthcare

The Group discussed this set of proposed action for the *Specialised Healthcare* pillar of the European Blueprint for Rare Diseases.

Actions selected as EU priorities:

2. Update and implement national rare disease plans with measurable objectives to improve access to specialised care, aligned with EU guidance and ERN integration, including financial incentives and stronger national engagement.

Add new action: Develop and implement generic care pathways for rare conditions grounded on the patient community needs and challenges.

ERN sustainability and integration (3 actions were merged under this heading)

16. Strengthen cross-border collaboration through ERNs, including sustainable funding, shared clinical pathways, and access to expertise

18. Ensure stable funding for ERNs at EU and Member State level.

19. Establish a formal ERN sustainability framework with clearer operational status, to secure protected expert time and accountability for care; data and research functions; training and strengthening Centres of Expertise capacities; enhancing ERNs competencies and services to better protect people living with a rare disease.

11. Strengthen registries and data linkage to support care quality, research and service planning

Actions selected as Global priorities:

11. Strengthen registries and data linkage to support care quality, research and service planning

Discussion

The discussion centred on how **integrated European Reference Networks (ERNs) could fully deliver on their potential**. Participants acknowledged the progress made to date, while emphasising that further action is needed to ensure all people living with a rare condition in the EU can benefit from this structure. It was also recognised that the extent to which individuals benefit from ERNs varies depending on their country of residence and the prevalence or incidence of their condition, and that each national health authority must determine how best to harness ERNs within its own health system.

Participants stressed that improving timely access to specialised healthcare requires stronger alignment between EU and national levels, to prevent people from falling through

gaps in the system. In particular, access to **cross-border healthcare** for people with rare conditions must be well integrated into national care pathways, whether through timely referral to an Expert Centre abroad or through ERN case discussion, with follow-up care at the local level properly coordinated with the treating Expert Centre.

More broadly, the group discussed the challenge of reaching **political agreement among Member States on the actions to be included in an EU Action Plan**. Since healthcare planning and delivery falls within exclusive national competence, country representatives must find common ground on the areas where they wish to deepen EU cooperation and define how that collaboration should be structured. While JARDIN may produce valuable recommendations and proofs of concept, Member States themselves must agree on where and how to move forward.

Notably, the group did not prioritise the two measures that received the highest number of votes in the Task Force Survey:

- **Awareness beyond specialists:** improving early recognition of rare diseases among non-specialist healthcare professionals;
- **Access to highly specialised treatments:** exploring the potential benefits, challenges, and feasibility of EU-level approaches to support cross-border access to ultra-rare and highly specialised treatments.

Both actions were discussed, each championed by one or two participants, but neither reached consensus for prioritisation. Equally, the group agreed not to discard actions that had received no votes (such as EU collaboration on workforce planning), recognising their continued relevance. Overall, the full list was viewed not as competing priorities but as a sequence of actions deliverable at different stages — some in the short term, others over a longer horizon.

Based on the agreed prioritisation criteria, the group reached swift and strong consensus on two actions:

- **National rare disease plans:** updating and implementing national plans with measurable objectives to improve access to specialised care, aligned with EU guidance and ERN integration, and supported by financial incentives and stronger national engagement
- **Registries and data use:** strengthening registries and data linkage to support care quality, research, and service planning – this action was also selected for the Global Action Plan, recognising the importance and power that can stem from data linkage and registries to enable access to quality care.

A sense of urgency around **embedding ERNs more firmly within national health systems** also drove agreement on a number of ERN-related actions. Well-established and

sustainable ERNs were seen as a prerequisite for enabling many of the other actions on the list.

Finally, participants agreed on the need for **clearer national care pathways** to guide patients from the very start of their journey. While this is an ongoing process that each country must shape according to its own context, the exchange of good practices was seen as a valuable lever. JARDIN recommendations are expected to provide further guidance in this area. This discussion prompted the group to define a new action, not previously listed, focused on developing and implementing **generic care pathways for rare conditions**, grounded in the needs and lived experiences of patient communities.



Overall, participants agreed on the need to **strengthen the integration of ERNs within national health systems and improve coordination** between EU and national levels to ensure better access to specialised care for people living with rare conditions. Strong consensus emerged around **national rare disease plans, registries and data use, and the development of clearer care pathways**, while recognising that progress will require a phased approach and sustained Member State commitment.

List of people registered for the discussion:

First Name	Surname	Affiliation
Ines	Hernando	EURORDIS (Rare Diseases Europe) - FACILITATOR
Anja	Helm	EURORDIS (Rare Diseases Europe) - Notetaker
Anna	Arellanesová	ČAVO (Czech Association for Rare Diseases)
Katri	Asikainen	HARSO ry (Finnish Alliance for Rare Diseases)
Janet	Bloor	Rare Disease Patient Advocate / JLB Law
Isabella	Brambilla	Dravet Italia Onlus
Alain	Coheur	Solidaris (Belgian Health Insurance Fund)
Natalie	Frankish	Genetic Alliance UK
Marta	Garcia Manrique	Servier
Cecilia	Gunnarsson	Region Östergötland / Linköping University Hospital

First Name	Surname	Affiliation
Victoria	Hedley	Newcastle University (Rare Disease Policy Research)
Alexandra	Keßmeyer	GBS CIDP Initiative Deutschland e.V.
Kris	Kusidlo	Sanfilippo Initiative / Global Childhood Dementia Consensus Project
Oleg	Kvlivdze	Georgian Foundation for Genetic and Rare Diseases (GeRaD)
Maria	La Forgia	Rare Disease Patient Representative / Clinical Researcher
Nawel	Lalout	World Duchenne Organization
Anne-Sophie	Lapointe	French Ministry of Health (National Plan for Rare Diseases)
Joyce	Loridan	Solidaris (Belgian Health Insurance Fund)
Eva	Motyčková	Rett Community Czechia (Rett-CZ)
Mari	Murel	Amsterdam UMC (Amsterdam University Medical Centers)
Charlotte	Rodwell	Orphanet / INSERM
Claas	Röhl	NF Kinder (Neurofibromatosis Association Austria)
Elisa	Rozzi	Emilia-Romagna Region (Rare Disease Network Coordination)
Josef	Schepers	University Medical Center Göttingen (UMG)
Jan	Swiderski	Kyowa Kirin
Enrique	Terol	Permanent Representation of Spain to the EU

6. Optimising the Data Ecosystem

The Group discussed this set of proposed action for the *Data* pillar of the European Blueprint for Rare Diseases.

Actions selected as EU priorities:

1. Integrate a dedicated rare disease module into the EHDS (European Health Data Space) with harmonised minimum datasets across Member States and legal frameworks facilitated for primary and secondary data use. This data infrastructure and registries shall follow FAIR and ERDRI standards and favour decentralised infrastructures with limited centralisation for specific research purposes.
2. Require the systematic use of OrphaCodes across health systems and datasets to improve visibility and comparability.
9. Support Member States in strengthening and implementing EHR systems to capture structured rare disease relevant data, ensuring compatibility with ERN registries, OrphaCodes, and EHDS.

Actions selected as Global priorities:

8. Ensure sustainable funding and quality management for national and EU registries, including integration of patient reported data where relevant
2. Require the systematic use of OrphaCodes across health systems and datasets to improve visibility and comparability
12. Enable secure secondary use of health data and real-world evidence with robust governance and consent models

Ex equo

16. Support validated AI applications for case finding, decision support, and research based on high-quality datasets (WHO, 2025; Nordic Roadmap, 2025) and monitor AI performance and equity impacts, ensuring transparency and accountability

Discussion

The discussion demonstrated strong consensus that establishing a coherent and **interoperable rare disease data ecosystem is a foundational prerequisite** for improving diagnosis, care, research, policy development, and access to innovation. Across interventions, participants repeatedly stressed that the challenge is no longer the absence of data, but rather the **fragmentation, limited interoperability, and underutilisation** of existing datasets and infrastructures. A clear trend emerged towards viewing data systems not as technical tools in isolation, but as **strategic enablers** capable of supporting the entire rare disease ecosystem.

Within this context, the **integration of a dedicated rare disease module into the European Health Data Space (EHDS)** (Action 1) was widely regarded as one of the most important and transformative actions, particularly at EU level. Participants consistently described this action as the mechanism capable of bringing together coding, interoperability, governance, consent frameworks, registries, and secondary use of data under a common European framework. Several contributors emphasised the need for a clear political mandate to ensure that rare disease requirements are explicitly embedded within EHDS implementation and regulation. This action was seen as comprehensive of all the core requirements for building a rare diseases ecosystem by most of the participants.

A second dominant theme concerned the **systematic adoption and implementation of OrphaCodes** (Actions 2, 3 and 4). Across discussions, OrphaCodes were repeatedly described as the key that would unlock the visibility of rare disease patients within healthcare systems, enabling better epidemiological knowledge, more reliable data collection, and meaningful secondary use of health information. Participants highlighted that many countries already possess large amounts of health data, but these remain difficult to use because rare disease patients are not consistently identifiable within health information systems. The discussion evolved beyond simple adoption of OrphaCodes towards the need for **mandatory implementation, harmonised application** across healthcare settings, mapping with existing coding systems, and systematic capture across hospital, primary care and mortality datasets. Several contributors argued that action 3 (support mapping between OrphaCodes and other systems) and action 4 (promote systemic capture of rare diseases codes across data sets) are intrinsically linked and could be merged into a single action focused on harmonised implementation and systematic use of OrphaCodes across health systems and datasets. More broadly, there may be value in considering a closer integration of actions 2, 3 and 4 into a **coherent coding and interoperability package**, reflecting the way these issues were consistently discussed together throughout the workshop.

Interoperability and harmonisation emerged as another major priority. Participants repeatedly stressed that Europe should focus on connecting and strengthening existing infrastructures rather than creating entirely new systems. The need to harmonise registry standards and **define common minimum datasets** (Action 5) received strong support, particularly as a means of reducing duplication and improving comparability across countries. Contributors highlighted that quality and harmonisation must progress together, noting that minimum datasets must remain sufficiently robust to support research, care planning and policy decisions. Similarly, discussions around establishing a European rare disease data hub (Action 14) reflected a growing preference for **federated and interconnected approaches to data sharing**. Rather than advocating for a highly centralised repository, participants favoured creating a framework capable of linking existing registries, biobanks and clinical data services while **preserving local ownership and governance**. Given the strong overlap between the objectives of actions 5 and 14, there

may be merit in explicitly linking these actions, positioning the data hub as the operational infrastructure through which registry harmonisation and interoperability are achieved.

The importance of **sustainable funding** emerged as one of the strongest and most consistent messages across the workshop. Action 8 received substantial support, particularly at global level, with participants repeatedly emphasising that even the most ambitious policies cannot be implemented without long-term financial investment. Several contributors highlighted that registries and data infrastructures require years to develop and maintain, and that funding should therefore be viewed as an enabling mechanism underpinning all other actions rather than as a standalone objective. The discussion suggested that sustainable financing models should explicitly support interoperability initiatives, quality management, patient-reported outcomes, and long-term maintenance of registries and data platforms.

The workshop also highlighted growing recognition of the **importance of governance frameworks that enable secure and trusted secondary use of health data**. Action 12 was frequently identified as a priority, particularly because of its potential to support research, clinical trials, real-world evidence generation and international collaboration. Participants stressed that substantial amounts of data already exist but remain inaccessible due to governance barriers rather than technical limitations. The discussion repeatedly emphasised the need to build on existing infrastructures and previous policy efforts rather than creating entirely new systems. Several speakers framed the urgency of improving data sharing not only as a technical challenge but also as a societal issue, arguing that delays in data sharing can have direct consequences for patients' lives.

Patient-centred governance emerged as a further cross-cutting theme. Participants stressed that patients should not only contribute data but should also retain meaningful control over how it is used and benefit from the knowledge generated through research and innovation. Action 18 received strong support, particularly in relation to dynamic consent models, transparency regarding data use, return of clinically relevant findings, and stronger patient ownership of data. Discussions frequently linked Action 18 with Action 12, suggesting that both actions contribute to a broader framework for trustworthy, patient-centred governance of rare disease data. A closer alignment between these actions could therefore be considered to strengthen coherence and avoid duplication.

The role of **digital technologies and emerging innovation** was also discussed extensively. While participants recognised the transformative potential of electronic health records (Action 9), artificial intelligence (Action 16), and patient-reported outcome measures (Action 13), there was broad agreement that these tools can only deliver meaningful value when supported by **high-quality, interoperable datasets and robust governance arrangements**. Participants highlighted that AI is no longer a future consideration, noting that patients are already using AI tools to interpret medical information and support decision-making. This generated calls for **accelerated development of governance frameworks** capable of

ensuring transparency, accountability, performance monitoring and equity in AI applications. Similarly, participants highlighted the importance of developing common approaches to PROMs and quality-of-life measures, particularly to support comparative evidence generation and strengthen the secondary use of health data.

Finally, the discussion reinforced the need to **view data policy as a strategic investment in the future of rare disease care** rather than as a purely technical undertaking. Across interventions, there was strong agreement that the most effective actions are those that create the conditions for future progress by improving visibility, interoperability, sustainability and trust.



The overall recommendation was therefore to **prioritise foundational measures capable of enabling broader systemic change**, particularly the integration of rare disease requirements within the EHDS, the systematic implementation of OrphaCodes, harmonisation of registries and datasets, sustainable funding mechanisms, and governance frameworks that facilitate secure, ethical and patient-centred use of health data.

List of people registered for the discussion:

Name	Surname	Affiliation
Nora	Lazaro	EURORDIS (Rare Diseases Europe) - FACILITATOR
Gemma	Rodríguez	EURORDIS (Rare Diseases Europe) - Notetaker
Avril	Daly	Retina International, EURORDIS
Toon	Digneffe	Takeda Pharmaceuticals (Rare Diseases Division)
John	Gerbild	Rare Disease Patient Advocate / Consultant (Denmark)
Emma	Green	Cambridge Rare Disease Network
Zdenek	Gütter	Ministry of Health of the Czech Republic (Rare Disease Policy)
Gudrun Helga	Hardardottir	Einstök Börn (Rare Diseases Iceland)

Name	Surname	Affiliation
Joelle	Khraiche	CSL Behring (Rare Diseases Portfolio)
Anna	Kociszewska-Kowalczyk	Polish Association of People with Rare Diseases / Patient Advocate
Rebecca	Leary	Newcastle University (John Walton Muscular Dystrophy Research Centre)
Davide	Marchi	Vertex Pharmaceuticals
Monica	Mazzucato	Veneto Region Rare Diseases Registry / Istituto Superiore di Sanità (ISS)
Samantha	Parker	Italfarmaco Group (Rare Disease Clinical & Regulatory Affairs)
Gábor	Pogány	HUFERDIS / RIROSZ (Hungarian Federation for Rare and Congenital Diseases)
Ana	Rath	Orphanet / INSERM (Institut National de la Santé et de la Recherche Médicale)
Pedro	Rocha	European Rett Syndrome Association (ERSA)
Solange	Roumengous	AP-HP (Assistance Publique - Hôpitaux de Paris / Rare Diseases Coordination)
Ece	Soyer Demir	SMA Disease Fight Association Turkey (SMA Benimle Yürü Derneği)
Simona	Stiskalova	Platforma Nezabudnutí (Slovak Rare Disease Support Platform)
Daniel	Theisen	ALAN - Maladies Rares Luxembourg
Dylan	Thurgood	Oxford Internet Institute / Rare Disease Patient Data Research
Ingrid	Vallee	The Rory Belle Foundation (Rare Genetic Disorders Advocacy)
Pascale	Van Durme	Radiorg (Belgian Rare Diseases Federation)
Maartje	Van Roosmalen	Johnson & Johnson (Janssen Rare Disease Portfolio)
Dariusz	Żebrowski	Stop Duchenne Foundation Poland

Conclusion & Next Steps

The Blueprint Workshop marked an important milestone in community engagement, multi-stakeholder dialogue, and consensus-building on the future of rare disease policy in Europe. Across all thematic areas, participants demonstrated strong alignment on the need for a more coordinated and ambitious response to the challenges faced by people living with rare diseases.

Discussions reinforced the view that many of these challenges transcend national borders and require collective solutions. Participants highlighted the importance of stronger European cooperation to reduce inequalities, share expertise, and ensure that advances in diagnosis, care, research, treatment access, and data systems benefit all communities. They also recognised the global relevance of these priorities, with many of the proposed actions providing a foundation for international collaboration and the development of a future Global Rare Disease Action Plan.

While priorities differed across pillars, a clear message emerged: fragmented efforts are no longer sufficient. Stakeholders voiced strong support for a dedicated EU Action Plan on Rare Diseases to provide strategic direction, accelerate progress, and translate commitments into tangible improvements for patients and families. The workshop outcomes therefore not only inform the finalisation of the European Blueprint for Rare Diseases, but also reflect a **united call for coordinated action at both European and global levels.**

The reflections and recommendations from the breakout sessions will feed into the broader consultation process led by EURORDIS, alongside the findings of the Regional Survey and the strategic guidance of the Core Group.

As work continues to refine the Blueprint and define the key structural elements of a future EU Action Plan on Rare Diseases, a **joint webinar will be held on 18 September to present the advanced draft ahead of publication.**