EURORDIS Response:

Position on the EU Biotech Act

EURORDIS-Rare Diseases Europe feedback to the European Commission consultation: Biotech Act

October 2025

Contents

Summary

The EU Biotech Act offers a unique opportunity to accelerate rare disease innovation and bridge the "valley of death" between scientific discovery and patient access to new treatments and diagnostic tools. With the appropriate end-to-end approach to transforming scientific innovations into final products, the Act could shift Europe's fragmented rare disease landscape into a coordinated, patient-centred, and innovation-driven ecosystem. As part of a broader, coherent and more streamlined legal framework, it can serve as a cornerstone for progress. The table below summarises the key policy demands necessary to achieve these objectives within the scope of the Act and unlock the full potential of biotechnology for rare diseases across Europe.

Policy proposals	Objectives
1. Set up a systemic, predictable, reusable, and flexible regulatory pathway for **Platform Technologies**, guided by a sandbox-like principle. This pathway should facilitate the **reuse of platform data across multiple products**, with the aim of reducing the burden of repetitive data generation and fostering sound regulatory approvals. It should also ensure the **active involvement of relevant stakeholders** — including patients — in developing guidance to support innovation and the efficient advancement of such technologies[1].	**Build EU Capacity for Technology Platforms**
2. **Set up an EU-funded network including expert centres, infrastructures and manufacturing facilities for key technology platforms with modular GMP**, clear rules to enable public private partnerships and patient co-governance, and practical measures to balance access between large and small actors (e.g., open-access conditions for EU-funded platforms)[2].	
3. Facilitate access to **innovative and alternative funding models** i.a. public-private partnerships, venture capital and public equity for companies, and other actors, including non-for profits and academic spinoffs scaling up key platform technologies (e.g., mRNA, genome editing, AI-based tools).	
4. **Formally embed innovative clinical trial design tools and innovative methodologies** (including adaptive, Bayesian, and digital monitoring methods, n-of-1/n-of-few trial designs) into trial assessment, approval and other regulatory processes. Reference outputs from EU-funded projects (RealiseD, ASTERIX, InSPiRe) in official guidance documents.	**Clinical Trials: Modern and high-quality designs**
5. **Enhance guidance and confidence in flexible evidence approaches** (e.g., single-arm studies, NHS real-world data, and surrogate endpoints) to address concerns over their methodological robustness and data quality, which currently limit their adoption. Regulatory frameworks should clarify appropriate use, set transparent quality standards, and promote consistent evaluation across regulators and HTA bodies to enable credible, clinically justified use.	
6. **Create an EU Clinical Trials Support Desk for logistical and financial support** to high quality care centres to qualify as clinical trial sites.	

[1] While the negotiations between the co-legislator are ongoing and pending the adoption of the General Pharmaceutical Legislation, this pathway may need to be aligned with the provisions that the GPL will introduce.

[2] Models include the BGTC in the US (https://fnih.org/our-programs/accelerating-medicines-partnership-amp/bespoke-gene-therapy-consortium-bgtc/) , the DCRT in the Netherlands (https://www.rnatherapy.nl/) and the UK RTLP (Rare Therapies Launch Pad, https://www.raretherapieslaunchpad.com/)

7. **Leverage European Reference Networks (ERNs) as trial platforms** to support cross border recruitment and coordination among specialist centres.	**Clinical Trials: Facilitate Cross-border Trials**
8. **Implement the EU-X-CT recommendations**, a set of guidelines aimed at harmonising and facilitating cross-border clinical trial participation in Europe, addressing ethics, costs, liability, and awareness to make trials more accessible for patients across EU countries.	
9. **Set up a EU Research Info Hub, a centralised information portal on ongoing research activities**, including clinical trials, real-world studies, compassionate use/early access programs, and medical device research. This platform shall ensure that patients and the public can readily identify and access research opportunities relevant to their condition. It will also provide support, streamline and manage the associated administrative processes.	
10. **Develop standards for collecting real-world evidence** (RWE) and digital endpoints, leveraging tools and outcomes from prior EU projects (e.g., RealiseD, ASTERIX, InSPiRe).	**Clinical Trials: Inclusion of RWE**
11. **Develop guidelines for strengthening the generation and availability of high-quality RWE to support its effective use into EMA regulatory processes and HTA evaluations** throughout the therapy lifecycle, including wearable monitoring, remote data capture, and patient-reported outcomes, to complement trials and inform regulatory decisions.	
12. Establish a centralised **EU funding mechanism** designed to enable **access to ATMPs for ultra-rare diseases** and ensure that all eligible patients can access treatments, regardless of their country of residence or treatment location.	**Financial Sustainability of Innovation**
13. **Mandate systematic uptake of ORPHAcodes** across national and European registries, electronic health record and healthcare systems in general along with measures to ensure the necessary support for Member State enforcement and digital readiness.	**Maximising Data and AI potential**
14. **Create possibilities for access to AI Factories and supercomputing infrastructures** to enable biotech SMEs, academia and non-profits to leverage advanced computation in product development.	
15. **Embed patient organisations as equal partners** across the research and policy ecosystem, while **ensuring long-term core funding** for patient organisations so that they can effectively contribute to and help shape innovation in Biotech.	**Patients' involvement**

1. The Benefits of Boosting Rare Diseases Biotech

The EU Biotech Act offers a strategic opportunity to strengthen Europe's position in biotechnology by accelerating the development and delivery of innovative therapies for rare diseases—an area of critical unmet medical needs. EU rare disease initiatives—like ERDERA, ERNs, registries, and patient-led partnerships—exist but remain fragmented, hampered by regulatory misalignment, infrastructure gaps, limited funding, and weak integration into EU strategies. Developing a coherent EU-wide strategy, in terms of biotech and beyond, is essential to unify these efforts, optimize resource allocation, ensure interoperability of data and technologies, reduce duplication, and accelerate research and patient access to therapies.

Tracking one of the biggest unmet needs in EU healthcare

While rare diseases impact approximately 30 million people across Europe, approved treatments are only available for around 5%[3][4]. Even when a treatment exists, a survey from 2024 found that 24% of people living with a rare disease (PLWRD) were unable to access a needed treatment in their country, compared to 7% of the general population[5]. Despite progress, numerous unmet medical needs and untapped scientific potential remain unaddressed in the field of rare and ultra-rare diseases.

Boosting EU's Competitiveness in Biotech

Rare diseases, including rare genetic disorders, autoimmune diseases, and rare cancers, are at the **forefront of biotechnological innovation**, having driven advances in genomics, proteomics, gene editing, cell biology, viral vectors (e.g., mRNA COVID vaccines), biomarkers, bioinformatics, and therapeutic development.[6] Scientific breakthroughs in this area have catalysed the uptake of **genomics** and inspired **national initiatives** such as Plan France Médecine Génomique 2025 in France,[7] Germany's genomeDE – National Strategy for Genomic Medicine,[8] and the European 1+ Million Genomes (1+MG) initiative[9]. With its experience in collaborative infrastructure, cross-border registries, open science and a legacy of innovation, including in adaptive clinical trials, Europe is well-positioned to become a global leader in this area.

However, between 2014–2018, clinical trials grew 36% in North America, 28% in Asia, but only 2% in Europe.[10][11] In 2024, Europe continued to lag in early-phase trials for advanced therapy medicinal products (ATMPs)[12]. Targeted investment and tailored policies for rare diseases, from early diagnosis to innovative therapies and drug repurposing, can reverse this trend. The Draghi Report identified rare diseases as a smart area for **restoring competitiveness**[13] and the Rare

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[3] EURORDIS (2019) *Rare2030 Trends*. Available at: https://www.rare2030.eu/trends/

[4] Nguengang Wakap, S. et al. (2020) "Estimating cumulative point prevalence of rare diseases: analysis of the Orphanet database", European Journal of Human Genetics, 28, pp. 165–173. Available at: https://doi.org/10.1038/s41431-019-0508-0.

[5] Eurordis (2024). Access to treatment: unequal care for European rare disease patients. Available at: https://www.eurordis.org/publications/access-to-treatment/. EURORDIS (2024). Rare disease patients' opinion on the future of rare diseases. Available at: https://download2.eurordis.org/rare2030/RARE2030_survey_public_report.pdf.

[6] [6] Examples of innovative therapies in this area include (1) Glybera (alipogene tiparvovec), a gene therapy approved in Europe in 2012 for treating Lipoprotein lipase deficiency (LPLD), a very rare genetic disorder affecting fat metabolism; (2) Strimvelis, an ex vivo gene therapy approved in 2016 in Europe to treat ADA SCID (adenosine deaminase deficiency-severe combined immunodeficiency), an ultra rare immune disorder; and (3) Onpattro (patirisan), a RNA interference (RNAi) drug, approved in 2018 in Europe to treat hereditary TTR-mediated amyloidosis.

[7] https://pfmg2025.fr/

[8] https://www.bundesgesundheitsministerium.de/en/en/international/european-health-policy/genomde-en.html

[9] https://digital-strategy.ec.europa.eu/en/policies/1-million-genomes

[10] Alliance for Regenerative Medicine (2019) *Clinical Trials in Europe: Recent Trends in ATMP Development*. Washington, D.C.: Alliance for Regenerative Medicine. Available at: https://alliancerm.org/wp-content/uploads/2019/10/Trends-in-Clinical-Trials-2019-Final_Digital.pdf

[11] Alliance for Regenerative Medicine (2024) *Clinical Trials in Europe: Recent Trends in ATMP Development*. Washington, D.C.: Alliance for Regenerative Medicine. Available at: https://alliancerm.org/wp-content/uploads/2025/02/Trials-2024-Final.pdf

[12] EURORDIS (2024, September 9). *Health innovation among proposals in Draghi's competitiveness report*. EURORDIS-Rare Diseases Europe. Available at: https://www.eurordis.org/eurordis-responds-to-draghi-report/

[13] EURORDIS (2024, September 9). *Health innovation among proposals in Draghi's competitiveness report*. EURORDIS-Rare Diseases Europe. Available at: https://www.eurordis.org/eurordis-responds-to-draghi-report/

2030 Foresight Study (rare2030.eu) highlighted the importance of investing in patient-driven innovation both to advance social justice and strategic advantages[12].

Reforms envisaged in the Act are especially critical for Europe's biotech start-ups, SMEs, academia and non-profit organisations, which are driving innovation in rare disease research but hindered by fragmented regulations and limited access to risk-tolerant capital. In addition, the expected integration of digital innovation and biotechnologies, will also be crucial for new rare disease treatments, diagnosis and screening, where every dataset is valuable and individualised approaches are often required.

Advancing Public Health Objectives

While rare diseases impact a smaller patient population, the scientific and technological advancements in this area, frequently translate into progress in other areas, such as more frequent cancers, cardiovascular diseases, and vaccines. Indeed, the **rare disease ecosystem holds significant potential to serve as a 'sandbox' for prototyping and piloting novel approaches and emerging technologies, with the capacity to scale and benefit broader healthcare systems and society at large**.

Rare Diseases Biotech also offer the potential to advance public health efforts by promoting early detection and intervention using genomics for newborn screening[14], pharmacogenomics for tailored therapeutic approaches[15], and polygenic risk scores for the identification of high-risk individuals[16] for specific diseases. Embedding these innovations into care pathways will also strengthen EU health system resilience and response capacity.

The costs of doing nothing: missing a unique opportunity

Limited commercial incentives, due to small patient populations and complex development pathways, discourage private investment in rare diseases[17]. Public intervention is crucial to bridging this gap and addressing the societal and economic burden it engenders. A 2024 study across nine EU countries and 43 diseases estimated the annual **economic burden of rare diseases at €249.3 billion**, exceeding the €176.8 billion cost of cardiovascular diseases[18]. Without further action, the EU risks losing its competitiveness while enduring the high economic as well as social costs of rare diseases.

Rare Diseases Biotech: one of the highest EU-Added Value sectors

Member States cannot address thousands of rare diseases alone. Only coordinated EU action will allow the pooling of expertise, data, and resources, which are essential due to the rarity and the knowledge fragmentation surrounding individual conditions. Hence, rare diseases are a natural area with **very high EU added value**[19][20].

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[14] Kingsmore, S.F. et al. (2022) "A genome sequencing system for universal newborn screening, diagnosis, and precision medicine for severe genetic diseases", *The American Journal of Human Genetics*, 109(9), pp. 1605–1619. Available at: https://pubmed.ncbi.nlm.nih.gov/36007526/

[15] Oates, J.T. and Lopez, D. (2018) "Pharmacogenetics: An Important Part of Drug Development with A Focus on Its Application", *International Journal of Biomedical Investigations*, 1(2), p. 111. Available at: https://www.ncbi.nlm.nih.gov/pmc/articles/PMC7255432/

[16] Lu, T. et al. (2021) "Individuals with common diseases but with a low polygenic risk score could be prioritized for rare variant screening", *Genetics in Medicine*, 23(3), pp. 508–515. Available at: https://www.nature.com/articles/s41436-020-01007-7

[17] Vavassori, S. et al. (2024) "Unlocking the full potential of rare disease drug development: exploring the not-for-profit sector's contributions to drug development and access", *Frontiers in Pharmacology*, 15, 1441807. Available at: https://doi.org/10.3389/fphar.2024.1441807

[18] CRA (Charles River Associates) (2024) *The Economic Cost of Living with a Rare Disease Across Europe*. Available at: https://media.crai.com/wp-content/uploads/2024/10/28114611/CRA-Alexion-Quantifying-the-Burden-of-RD-in-Europe-Full-report-October2024.pdf

[19] European Commission (2008) *Rare diseases: Europe's challenges*. Communication from the Commission to the European Parliament, the Council, the European Economic and Social Committee and the Committee of the Regions (COM(2008) 679 final). Brussels, 11 November. Available at: https://eur-lex.europa.eu/legal-content/EN/TXT/?uri=CELEX:52008DC0679

[20] Council of the European Union (2009) Council Recommendation of 8 June 2009 on an action in the field of rare diseases. 2009/C 151/02. *Official Journal of the European Union*, C 151, 3 July, pp. 7–10. Available at: https://eur-lex.europa.eu/legal-content/GA/TXT/?uri=CELEX:32009H0703(02)

While many rare disease initiatives already exist at EU level, such as ERDERA (European Rare Disease Research Alliance, erdera.org), European Reference Networks (ERNs), data registries and patient-led partnerships, efforts remain fragmented across geographies and technologies, hindered by regulatory misalignment, infrastructure gaps, limited capital, and insufficient integration into broader EU strategies.
An ambitious Biotech Act, should therefore, be operationalised, where applicable, through an **EU Action Plan for Rare Diseases**[21] with clear, actionable and trackable goals (as proposed by EURORDIS and supported by the European Parliament, the European Economic and Social Committee and many Member States), which would be **required to help Europe reverse its declining global competitiveness in biotechnology**.

2. Flagship EURORDIS Recommendation: Build EU Capacity for Technology Platforms

COVID-19 taught us a lesson: if mRNA platform could go from sequence to vaccine in months, then having shared tools (like delivery vectors, data models, or editing enzymes) can significantly accelerate the R&D pathway. Platform technologies hold the potential to revolutionise the biotech sector. While their definition is still being discussed, including in the context of the Revision of the EU General Pharmaceutical Legislation,[22] **platform technologies are widely considered as** inherently versatile and rapidly adaptable to respond to emerging health threats or to address unmet medical needs.

Across Europe, the development of therapies for ultra-rare and individualised conditions remains fragmented, slow, and heavily dependent on *ad-hoc* academic or charitable initiatives. Promising scientific discoveries—such as patient-specific antisense oligonucleotides or modular viral vectors—often fail to progress beyond proof-of-concept because researchers and small developers lack access to Good Manufacturing Practices (GMP)-compliant manufacturing, regulatory expertise, and sustainable financing. Each new therapy must repeatedly overcome the same technical, regulatory, and clinical barriers, wasting time and public investment. While the United States and the United Kingdom are already building national infrastructures to standardise and accelerate platform therapies[23], the EU --with a few exceptions such as the DCRT in the Netherlands-- still lacks coordinated, locally based technology platforms capable of translating laboratory innovation into safe, reproducible treatments for patients. Without such platforms—anchored in well-established rare disease research groups connected with international research networks and supported by stable EU investment—Europe risks losing leadership in advanced biotechnologies and, most importantly, failing to deliver lifesaving innovation to people living with rare and ultra-rare diseases.

In funding and developing infrastructure and other facilities for Platform Technologies, it will be crucial for the EU to ensure practical measures to balance access between large and small actors (e.g., open-access conditions for EU-funded platforms).

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[21] https://download2.eurordis.org/rare2030/Rare2030_Action01_factsheet.pdf and https://download2.eurordis.org/rare2030/EURORDIS%20Action%20Plan%20Concept%20Note_Nov%2021_final.pdf

[22] In scientific literature, these technologies are described as foundational frameworks enabling faster, more efficient, and transferable innovation—exemplified by mRNA vaccine platforms or viral vector systems in gene therapy. The U.S. Food and Drug Administration considers that a platform technology "facilitates the development or manufacture of more than one drug through a standardized process." Food and Drug Administration (2024) Designating a Platform Technology for Use in Drug and Biological Product Development. Silver Spring, MD: U.S. FDA),

[23] Models include the BGTC in the US (https://fnih.org/our-programs/accelerating-medicines-partnership-amp/bespoke-gene-therapy-consortium-bgtc/) , the DCRT in the Netherlands (https://www.rnatherapy.nl/) and the UK RTLP (Rare Therapies Launch Pad, https://www.raretherapieslaunchpad.com/)

Proposed policy solutions

- Set up a systemic, predictable, reusable, and flexible regulatory pathway for **Platform Technologies**, guided by a sandbox-like principle. This pathway should facilitate the **reuse of platform data across multiple products**, with the aim of reducing the burden of repetitive data generation and fostering sound regulatory approvals. It should also ensure the **active involvement of relevant stakeholders** — including patients — in developing guidance to support innovation and the efficient advancement of such technologies[24].
- **Set up an EU-funded network including expert centres, infrastructures and manufacturing facilities for key technology platforms with modular GMP**, clear rules to enable public private partnerships and patient co-governance, and practical measures to balance access between large and small actors (e.g., open-access conditions for EU-funded platforms)[25].
- Facilitate access to **innovative and alternative funding models** i.a. public-private partnerships, venture capital and public equity for companies, and other actors, including non-for profits and academic spinoffs scaling up key platform technologies (e.g., mRNA, genome editing, AI-based tools).

3. Delivering on the EU Biotech Act: Innovation, Evidence, and Patient Access

Bringing innovative therapies for rare diseases from 'bench to bedside' remains one of the most critical challenges, with one or more “valleys of death” –where promising scientific discoveries fail to progress into approved treatments. Clinical trials are central to bridging this gap, but traditional trial designs frequently struggle to accommodate the small, heterogeneous populations characteristic of rare diseases, leading to delays, underpowered studies, or trials that are never initiated. By embracing modern, high-quality trial designs, cross-border recruitment strategies, real-world evidence integration, and advanced data-driven approaches, the EU has a unique opportunity to reduce this translational bottleneck. By complementing the provisions of the Clinical Trials Regulation, the Biotech Act shall establish measures that can accelerate therapy development, improve patient access, and foster a more inclusive, patient-centred ecosystem for rare disease innovation.

Clinical Trials: Modern and high-quality designs

The EU Biotech Act provides an important opportunity to enhance the regulatory framework for rare diseases by promoting modern clinical trial designs. Traditional methodologies often struggle to meet the needs of small, heterogeneous patient populations, resulting in delays in and failed therapy development. Incorporating innovative approaches—such as adaptive, platform, and decentralised trials—can streamline drug development, reduce patient burden, and accelerate access to treatments.

Proposed policy solutions

- **Formally embed innovative clinical trial design tools and innovative methodologies** (including adaptive, Bayesian, and digital monitoring methods, n-of-1/n-of-few trial designs) into trial assessment, approval and other regulatory processes. Reference

[24] While the negotiations between the co-legislator are ongoing and pending the adoption of the General Pharmaceutical Legislation, this pathway may need to be aligned with the provisions that the GPL will introduce.

[25] Models include the BGTC in the US (https://fnih.org/our-programs/accelerating-medicines-partnership-amp/bespoke-gene-therapy-consortium-bgtc/) , the DCRT in the Netherlands (https://www.rnatherapy.nl/) and the UK RTLP (Rare Therapies Launch Pad, https://www.raretherapieslaunchpad.com/)

outputs from EU-funded projects (RealiseD, ASTERIX, InSPiRe) in official guidance documents.

- **Enhance guidance and confidence in flexible evidence approaches** (e.g., single-arm studies, NHS real-world data, and surrogate endpoints) to address concerns over their methodological robustness and data quality, which currently limit their adoption. Regulatory frameworks should clarify appropriate use, set transparent quality standards, and promote consistent evaluation across regulators and HTA bodies to enable credible, clinically justified use.
- **Create an EU Clinical Trials Support Desk for logistical and financial support** to high quality care centres to qualify as clinical trial sites.

Clinical Trials: Facilitate Cross-Border

Modernising and harmonising clinical trial processes across Member States, particularly for small and hard-to-reach populations, is essential to unlock the full potential of biotech innovation in Europe. As new therapies increasingly rely on novel mechanisms of action or target small populations, traditional clinical trials alone may not capture the full picture of safety, efficacy, or long-term value.

Rare disease trials are hindered by the difficulty of recruiting enough patients across dispersed populations[26]. Cross-border trials, which are essential for rare diseases, remain rare due to national regulatory and logistical hurdles. National differences in ethics approval procedures, regulatory requirements, language, and logistics make multinational trials particularly burdensome and costly[27] and existing EU legislation on cross-border healthcare does not specifically address participation in clinical trials. A study on cross-border access to clinical trials found that while 92% of survey respondents saw the ability to access trials abroad as necessary, actual participation across borders remains rare[28]. These issues disproportionately affect smaller sponsors and contribute to delays, underpowered studies, or trials that are never launched[29].

Proposed policy solutions

- **Leverage European Reference Networks (ERNs) as trial platforms** to support cross border recruitment and coordination among specialist centres.
- **Implement the EU-X-CT recommendations**, a set of guidelines aimed at harmonising and facilitating cross-border clinical trial participation in Europe, addressing ethics, costs, liability, and awareness to make trials more accessible for patients across EU countries.
- **Set up a EU Research Info Hub, a centralised information portal on ongoing research activities**, including clinical trials, real-world studies, compassionate use/early access programs, and medical device research. This platform shall ensure that patients and the public can readily identify and access research opportunities relevant to their condition. It will also provide support, streamline and manage the associated administrative processes.

[26] Day, S. et al. (2018) "Recommendations for the design of small population clinical trials", *Orphanet Journal of Rare Diseases*, 13(1), 195. Available at: https://doi.org/10.1186/s13023-018-0931-2
[27] Lalova, T. et al. (2020) "Cross-border access to clinical trials in the EU: exploratory study on needs and reality", *Frontiers in Medicine*, 7, 585722. Available at: https://doi.org/10.3389/fmed.2020.585722
[28] Ibid
[29] del Álamo, M. et al. (2022) "Identifying obstacles hindering the conduct of academic-sponsored trials for drug repurposing on rare diseases: an analysis of six use cases", *Trials*, 23, 783. Available at: https://doi.org/10.1186/s13063-022-06713-y

Clinical Trials: Integration of Real-World Evidence

Real World Evidence is becoming a cornerstone of innovation in the life sciences sector: by capturing patient experiences outside traditional clinical trials, it provides a broader, more accurate picture of how treatments work in real life. Incorporating RWE into regulatory processes can complement clinical trial data, improve decision-making, and accelerate access to therapies. Incorporating these insights ensures that the Biotech Act fosters a regulatory environment that is more inclusive, responsive to patient experiences, and supportive of a patient-centred, efficient research ecosystem.

Proposed policy solutions

- **Develop standards for collecting real-world evidence** (RWE) and digital endpoints, leveraging tools and outcomes from prior EU projects (e.g., RealiseD, ASTERIX, InSPiRe).
- **Develop guidelines for strengthening the generation and availability of high-quality RWE to support its effective use into EMA regulatory processes and HTA evaluations** throughout the therapy lifecycle, including wearable monitoring, remote data capture, and patient-reported outcomes, to complement trials and inform regulatory decisions.

Financial Sustainability of Innovation

Within the framework of the Life Science Strategy and the EU Biotech Act, establishing a centralised funding mechanism for advanced therapy medicinal products (ATMPs) targeting ultra-rare diseases represents a critical step towards both equity and financial sustainability in healthcare. The Act's overarching objective of fostering innovation in the biotechnology sector is complemented by ensuring that patients across all Member States can access life-changing therapies, irrespective of national budgetary constraints or geographic location. By pooling resources and sharing financial risk at the EU level, the mechanism would reduce disparities in treatment availability, incentivise continued development of high-cost, high-impact therapies, and provide a predictable market environment for innovators.

Proposed policy solutions

- **Establish a centralised EU funding mechanism** designed to enable access to ATMPs for ultra-rare diseases and ensure that all eligible patients can access treatments, regardless of their country of residence or treatment location.

Maximising Data and AI potential

The ability to collect, structure, share, and analyse data across rare disease research and care is essential – but remains inconsistent and underutilised. A major barrier is the incomplete uptake of ORPHAcodes, the rare disease-specific coding system developed by Orphanet, which is still not systematically adopted across healthcare systems, registries, regulatory submissions, and health technology assessment (HTA) processes. And while systematic ORPHA code uptake is crucial, its realisation ultimately depends on Member State enforcement and digital readiness at national level.

This undermines interoperability and the coordination of EU-wide efforts. Meanwhile, the potential of data-driven science – including AI and machine learning – remains largely untapped, hindered by limited data quality, lack of labelling, weak incentives for open science and inadequate research reproducibility due to heterogeneous data sources and standards. Rare disease patients overwhelmingly support data sharing when it serves public-interest goals like improving diagnosis, understanding disease mechanisms, and developing treatments. They also

expect meaningful control, clear information on how their data is used, and strong safeguards[30]. Building trust through transparency, accountability, and engagement is essential to the success of data-driven innovation.

Proposed policy solutions

- **Mandate systematic uptake of ORPHAcodes** across national and European registries, electronic health record and healthcare systems in general along with measures to ensure the necessary support for Member State enforcement and digital readiness.
- **Create possibilities for access to AI Factories and supercomputing infrastructures to enable biotech SMEs**, academia and non-profits to leverage advanced computation in product development.

Patients as Equal Partners in Innovation and Policy

Patients' lived experience offers vital insights into coping with rare diseases and identifying unmet needs – essential for shaping relevant, impactful research and for the development of new therapies and diagnostics. Yet, despite growing recognition, patients and patient organisations remain underrepresented in the governance and design of research and their contributions are still underfunded, with limited resources to support patient-led initiatives or enable sustained engagement. A Rare 2030 survey found that 95% of patient representatives were willing to be actively involved in research projects as partners or co-investigators[31]. Yet to do so, most indicated they would need financial support (80%), human resources (69%), and skills development (60%)[32].

Proposed policy solutions

- **Embed patient organisations as equal partners** across the research and policy ecosystem, while **ensuring long-term core funding** for patient organisations so that they can effectively contribute to and help shape innovation in Biotech.

4. Creating the Conditions for Success: Enabling Actions Beyond the Biotech Act

To fully harness the potential of biotechnology in Europe—particularly to address unmet medical needs in rare diseases— the regulatory framework must evolve to better foster innovation, reduce fragmentation, and enable timely patient access to innovative solutions. This evolution shall culminate in a coherent, streamlined and innovation-friendly legislative and regulatory ecosystem, with the Biotech Act serving as a key, foundation pillar.

Achieving this vision requires a series of complementary measures that extend beyond the immediate scope of the Biotech Act. Together, they will create the enabling conditions for the Act's objectives to be fully realised and sustained over time.

Complementary Measures

Review the Clinical Trials Regulation to create an EU environment capable of supporting multi-country CTs and modernising the design of CTs to reflect the needs of modular technologies and small patient populations. This should include, among other things

[30] Courbier, S. et al. (2019) "Share and protect our health data: an evidence based approach to rare disease patients' perspectives on data sharing and data protection", *Orphanet Journal of Rare Diseases*, 14, 175. Available at: https://doi.org/10.1186/s13023-019-1123-4

[31] EURORDIS (2021) *Rare 2030 survey: Rare disease patients' opinion on the future of rare diseases*. Paris: EURORDIS-Rare Diseases Europe. Available at: https://download2.eurordis.org/rbv/rare2030survey/reports/RARE2030_survey_public_report_en.pdf.

[32] EJP RD (2021) *Short guide on patient partnerships in rare disease research projects*. European Joint Programme on Rare Diseases. Available at: https://www.ejprarediseases.org/wp-content/uploads/2021/03/SHORT-GUIDE-ON-PATIENT-PARTNERSHIPS-IN-RARE-DISEASE-RESEARCH-PROJECTS.pdf

- Improving the coordination between ethic committees and streamlining approval timelines for CTs to reduce burden on authorities and sponsors, and
- Including a **Formal Patient Consultation Mechanism for trials** (especially in rare diseases or other areas of high unmet need) for patients to bring their concerns about a trial, and reflecting the needs of modular technologies and small patient populations.

Advance the goals of ACT-EU by reducing red tape for multinational and academic-led trials. The project set ambitious objectives that should be further reflected in the Biotech Act.

- Improve the clinical research environment in Europe while maintaining strong participant protection and data quality.
- Strengthen coordination, ethical oversight and support for large multinational trials.
- Reduce administrative burden and increase efficiency.
- Prioritise trials for unmet medical needs, rare diseases and public health emergencies, supporting both academic and small industry sponsors.
- Provide coordinated scientific advice to enhance trial impact and support medicine authorisation and access.
- Promote inclusive, patient-centred medicines development.
- Present a unified European position internationally on clinical trials.
- Build skills and capacity through research collaboration and training.

Review the EU legal framework for cross-border healthcare—covering Directive 2011/24/EU and Regulation (EC) No 883/2004—to harmonise access and certification procedures (e.g. S2/F2 frameworks). The current dual system has created bureaucratic complexity and lacks a clear pathway for accessing transformative treatments such as ATMPs, posing major administrative and logistical challenges for healthcare systems. Even without legislative revision, minimum actions should include:

- Developing EU-wide guidance for national authorities on navigating the dual system, with clearly defined roles for insurers and payers.
- Issuing European Commission guidance on the applicable legal instruments for ATMPs following EMA centralised approval, particularly when treatments are delivered in a limited number of specialised centres.

In synergy with the **EU Life Sciences Strategy** (LSS):

- **Continue supporting and ensure the long-term sustainability of ERDERA**, the Rare Disease Research Partnership, and the infrastructures it has established – such as the Acceleration Hub – while further integrating these with EU and national research ecosystems.
- **Commit to long-term investment in ERNs and their Centres of Expertise** as they represent hubs of excellence for care, and harness their potential for research, and innovation. As a pre-condition, it is essential to enable and simplify ERN **collaboration with the private sector**, enabling ERNs to source funding from private actors who can support the federation of existing data registries, create sub-registries, and capture rich data to advance knowledge and stimulate research.
- **Strengthen public–private partnerships** involving academia, industry, regulators, and patient groups, for instance to share resources -such as molecular libraries- and technical expertise for rare disease drug development.
- **Encourage and fund longitudinal registries** capturing comprehensive patient histories over time, which can provide real-world insights for better treatment personalisation.
- **Ensure Equitable Digital Access for Patients** by promoting policies that actively bridge the digital divide to ensure that rare disease patients in rural, remote, and marginalised

communities can fully benefit from telemedicine, digital health tools, and data-sharing platforms. This requires investment in broadband and mobile infrastructure, affordable or subsidised devices and secure digital platforms for patients and caregivers, and accessible digital literacy support tailored to different levels of technological familiarity and language needs. It will be crucial to ensure that further digitalisation closes rather than increases existing access gaps.