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DRAFT REPORT

with recommendations to the Commission on a European Union rare disease action plan
(2025/2130(INL))

Committee on Public Health

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(Initiative – Rule 47 of the Rules of Procedure)

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MOTION FOR A EUROPEAN PARLIAMENT RESOLUTION

with recommendations to the Commission on a European Union rare disease action plan (2025/2130(INL))

The European Parliament,

– having regard to Article 225 of the Treaty on the Functioning of the European Union,

– having regard to Articles 16, 114 and 168 of the Treaty on the Functioning of the European Union,

– having regard to the Charter of Fundamental Rights of the European Union,

– having regard to the Council of Europe Convention for the Protection of Human Rights and Fundamental Freedoms,

– having regard to the Council of Europe Convention of 4 April 1997 for the Protection of Human Rights and Dignity of the Human Being with regard to the Application of Biology and Medicine,

– having regard to Regulation (EU) No 536/2014 of the European Parliament and of the Council of 16 April 2014 on clinical trials on medicinal products for human use, and repealing Directive 2001/20/EC[1],

– having regard to Regulation (EU) 2021/2282 of the European Parliament and of the Council of 15 December 2021 on health technology assessment and amending Directive 2011/24/EU[2],

– having regard to Regulation (EU) 2023/2854 of the European Parliament and of the Council of 13 December 2023 on harmonised rules on fair access to and use of data and amending Regulation (EU) 2017/2394 and Directive (EU) 2020/1828 (Data Act)[3],

– having regard to Regulation (EU) 2024/1938 of the European Parliament and of the Council of 13 June 2024 on standards of quality and safety for substances of human origin intended for human application and repealing Directives 2002/98/EC and 2004/23/EC[4],

– having regard to Regulation (EU) 2025/327 of the European Parliament and of the Council of 11 February 2025 on the European Health Data Space and amending Directive 2011/24/EU and Regulation (EU) 2024/2847[5],

– having regard to Directive 2011/24/EU of the European Parliament and of the Council

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1 OJ L 158, 27.5.2014, p. 1, ELI: http://data.europa.eu/eli/reg/2014/536/oj.
2 OJ L 458, 22.12.2021, p. 1, ELI: http://data.europa.eu/eli/reg/2021/2282/oj.
3 OJ L, 2023/2854, 22.12.2023, ELI: http://data.europa.eu/eli/reg/2023/2854/oj.
4 OJ L, 2024/1938, 17.7.2024, ELI: http://data.europa.eu/eli/reg/2024/1938/oj.
5 OJ L, 2025/327, 5.3.2025, ELI: http://data.europa.eu/eli/reg/2025/327/oj .

on the application of patients' rights in cross-border healthcare[6],

– having regard to the proposal for a regulation of the European Parliament and of the Council laying down Union procedures for the authorisation and supervision of medicinal products for human use and establishing rules governing the European Medicines Agency, amending Regulation (EC) No 1394/2007 and Regulation (EU) No 536/2014 and repealing Regulation (EC) No 726/2004, Regulation (EC) No 141/2000 and Regulation (EC) No 1901/2006 (COM(2023)0193),

– having regard to the proposal for a directive of the European Parliament and of the Council on the Union code relating to medicinal products for human use, and repealing Directive 2001/83/EC and Directive 2009/35/EC (COM(2023)0192),

– having regard to conclusion of the Council of 29 May 2024 on the Future of the European Health Union: A Europe that cares, prepares and protects,

– having regard to the Commission communication of 4 March 2021 on the European Pillar of Social Rights Action Plan,

– having regard to the conclusions and recommendations of 29 November 2024 of the European Economic and Social Committee conference for an EU commitment to tackling rare diseases,

– having regard to the report of November 2023 of the European Parliamentary Research Service, the Scientific and Technological Options Assessment panel, Improving access to medicines and promoting pharmaceutical innovation,

– having regard to the European added value assessment of February 2026 carried out by the European Parliamentary Research Service on an EU rare disease action plan,

– having regard to the opinion of the Committee on Legal Affairs on the proposed legal basis,

– having regard to Rules 47 and 55 of its Rules of Procedure,

– having regard to the report of the Committee on Public Health (A[10-0000/2026]),

A. whereas rare diseases affect an estimated 27 to 36 million people in the Union and the number of recognised conditions continues to grow as science advances;

B. whereas of the estimated 6 000-8 000 rare diseases identified, even a minimal level of scientific knowledge only exists for fewer than 1 000 of them and about 95 % lack approved treatment, contributing to unmet medical needs[7];

C. whereas around 80 % of rare diseases have a genetic origin, and the symptoms can overlap with common conditions, further complicating early detection and diagnosis;

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[6] OJ L 88, 4.4.2011, p. 45, ELI: http://data.europa.eu/eli/dir/2011/24/oj.

[7] Rare diseases – strengthening EU action - https://www.europarl.europa.eu/RegData/etudes/ATAG/2025/779215/EPRS_ATA(2025)779215_EN.pdf

D. whereas a public consultation conducted by the European Parliament in March 2025 gathered the views of more than 4 000 persons confirming widespread support for the development of a comprehensive European Rare Disease Plan to strengthen coordination and address these persistent challenges;

E. whereas existing Union action on rare diseases has delivered important tools, but significant disparities continue to exist between and within Member States;

F. whereas the European partnership on research on rare diseases represents a significant collaborative effort, bringing together partners from across the Union to drive research in prevention, diagnosis and treatment;

G. whereas civil society organisations have repeatedly called for a comprehensive Union action plan on rare diseases, reaffirming that fragmented national approaches limit effectiveness and equity in care;

H. whereas the European Parliament has called for a European plan in rare and neglected diseases in its resolution of 10 July 2020 on the EU's public health strategy post-COVID-19[8];

I. whereas the European Economic and Social Committee encouraged further action to establish a coherent framework that guides national policies and encourages collaborative action beyond European Reference Networks (ERNs);

J. whereas a coherent Union rare disease action plan including the introduction of legislation for a rare diseases action framework could address gaps and deliver better coordination and equity across the care pathway;

K. whereas registries and interoperable data exchange were viewed as crucial to improving understanding of rare diseases;

Rare diseases plan framework

1. Calls on the Commission and Member States to develop a comprehensive Union rare disease action plan that addresses the full rare disease care pathway;

2. Urges the creation of mechanisms to improve access and affordability of therapies, including targeted support for ultra-rare conditions and advanced therapies;

3. Encourages strategic Union-level research and innovation coordination, aligning research agendas, pooling resources for shared infrastructures;

4. Calls for comprehensive frameworks for patient and family support and Union-wide evidence generation on socio-economic impacts, with attention to gender and caregiving inequalities;

5. Stresses the need to strengthen governance and cross-border coordination by enhancing the sustainability, integration, geographical coverage and referral pathways of ERNs,

[8] OJ C 371, 15.9.2021, p. 102.

and exploring mechanisms for stable financing and legal status of the ERNs;

Diagnosis, screening and early detection

6. Calls on the Commission and Member States to accelerate efforts to reduce diagnostic delay for rare diseases;

7. Urges the development of common principles for the screening of newborns and early detection to tackle social and territorial inequality across the Union;

8. Encourages the creation of a Union coordination mechanism to align and strengthen screening programmes and diagnostic pathways across Europe;

9. Supports greater use of genomic and advanced diagnostic technologies, telemedicine and digital tools, facilitating cross-border diagnostic;

Access to treatment, affordability and innovation

10. Stresses the need for reinforced Union-level coordination on health technology assessment and evidence evaluation for rare disease treatments;

11. Calls on Member States to support voluntary joint clinical evidence generation, joint procurement and solidarity-based approaches;

12. Encourages tailored regulatory pathways and incentives for ultra-rare conditions and advanced therapies;

13. Supports greater transparency in treatment availability and pricing components where feasible;

14. Calls for systematic monitoring of inequalities in access, reimbursement timelines and availability of authorised treatments across Member States;

15. Encourages the exploration and piloting of sustainable funding and payment models for advanced therapy medicinal products, including outcome-based and risk-sharing arrangements;

Research, development and innovation ecosystem

16. Calls for a Union-wide rare disease research strategy aligned with Horizon Europe funding;

17. Encourages the pooling of research infrastructures and biobanks;

18. Supports coordinated research agendas to avoid duplication;

19. Calls for increased patient involvement in research governance;

20. Encourages public-private partnerships focused on unmet needs;

21. Supports Union funding for translational and clinical research;

22. Calls for improved cross-border clinical trial coordination;

23. Requests measurable targets for research collaboration and output;

Health data, registries and digital infrastructure

24. Calls for full interoperability of rare disease registries and further development of data sharing and rare disease registry infrastructures;

25. Supports the integration of rare disease data into the European Health Data Space established by Regulation (EU) 2025/327 as an essential step of the process of its implementation;

26. Encourages the systematic use of standardised coding systems;

27. Calls for a federated European rare disease data network as a step to be incorporated in the deployment of the European Health Data Space;

28. Encourages secure cross-border data sharing respecting privacy rules;

Patient support, social inclusion and care pathways

29. Calls for Union guidance on integrated, multidisciplinary care pathways;

30. Encourages recognition of the socio-economic impact on families and carers;

31. Supports harmonised principles for disability assessment based on functional impact;

32. Calls for improved cross-border continuity of care;

Governance, Member States coordination and cross-border cooperation

33. Calls for a comprehensive Union rare disease action plan with clear milestones setting a strategic roadmap for all pillars of rare disease policy;

34. Encourages the reinforcement and sustainable financing of ERNs as central engines of cross-border clinical cooperation;

35. Supports reinforced ERNs infrastructure to enhance and regulatory strength to ensure the Commission’s coordination and leadership;

36. Encourages the development of Union-level benchmarks and scoreboards;

37. Calls for evaluation mechanisms to measure socio-economic impact and cost savings;

38. Instructs its President to forward this resolution and the accompanying recommendations to the Commission and the Council.

ANNEX TO THE MOTION FOR A RESOLUTION: RECOMMENDATIONS AS TO THE CONTENT OF THE PROPOSAL REQUESTED

A. PRINCIPLES AND AIMS OF THE PROPOSAL

Recommendation 1 (on the form and minimum content of the instrument to be adopted)

The European Parliament considers that the legislative act to be adopted should establish a comprehensive and binding European rare disease action framework in order to address fragmentation, inequalities and inefficiencies in diagnosis, treatment, research coordination and data infrastructure across the Union.

The European Parliament considers that the legislative act to be adopted should:

– establish a coherent Union framework covering early diagnosis, screening coordination, access to medicinal products and advanced therapies, research cooperation, interoperable data infrastructures, integrated care pathways, governance mechanisms and monitoring systems;

– lay down common principles and coordination obligations to ensure interoperability of rare disease registries, compatibility with the European Health Data Space and structured exchange of real-world evidence across Member States;

– strengthen and structurally reinforce the legal base of the European Reference Networks (ERNs) and the leadership of the Commission including sustainable Union financing and clearer governance mechanisms;

– introduce a Union-level monitoring and reporting mechanism, including common indicators and measurable targets, to assess progress in reducing diagnostic delay, improving equitable access to treatment and enhancing cross-border cooperation;

– provide mechanisms to promote the use of cross-border pathways for advanced therapy medicinal products, and the scaling-up of its developments;

– promote the openness of platform technology master files, enabling faster and further research while lowering costs of developments through public fund development partnerships;

– ensure structured, integrated and regular consultation of patient organisations, healthcare professionals and civil society in the governance and evaluation of the framework;

– define a clear review clause and periodic evaluation mechanism to assess effectiveness, proportionality and European added value.

The European Parliament considers that the appropriate legal instrument is a Regulation, in order to guarantee uniform application of coordination standards, interoperability requirements and governance mechanisms across the Union, thereby preventing further legal and operational fragmentation that could arise from divergent national transposition.

The European Parliament considers that the Regulation should respect the competences of the Member States concerning the organisation and delivery of healthcare, while establishing

binding cooperation mechanisms and minimum coordination requirements necessary to achieve European added value.

Recommendation 2 (on the objectives, guiding principles and structural architecture of the framework)

The European Parliament considers that the legislative act to be adopted should ensure coherence between policy objectives and implementation mechanisms, based on the principle that the scale and cross-border nature of rare diseases require coordinated Union action in accordance with subsidiarity and proportionality.

For those reasons, the European Parliament considers that the framework should:

– be structured around six interdependent pillars: early and accurate diagnosis; equitable access to treatments and advanced therapies; research coordination and innovation; interoperable data and digital infrastructure; integrated and patient-centred care pathways; and governance, monitoring and financing;

– ensure that the place where expertise is located and the place where the patient resides do not determine the level of access to diagnosis or treatment;

– reduce diagnostic delay through coordinated principles on screening of newborns, voluntary convergence of genomic testing standards and strengthened cross-border referral mechanisms to promote social equality and tackle the social disadvantages for children due to the unavailability of early diagnosis and screening programmes;

– enhance equitable access to orphan medicinal products and advanced therapies through coordinated evidence generation, voluntary joint procurement mechanisms and strengthened cooperation in health technology assessment;

– integrate rare disease registries and data infrastructures within the European Health Data Space in a manner that ensures interoperability, cybersecurity and full compliance with Union data protection legislation;

– strengthen ERNs as hubs for clinical expertise, data exchange, training and research coordination, ensuring sustainable financing and integration with national systems;

– promote cross-border continuity of care and multidisciplinary support services, including psychosocial and socio-economic support for patients and caregivers;

– establish and coordinate the development of a hospital exemption cross-border exchange scheme;

– define measurable Union-level targets, including objectives relating to maximum average diagnostic delay, rare disease registry interoperability coverage and equitable availability of authorised therapies across Member States.

The European Parliament considers that the structural design of the framework should be outcome-oriented, measurable and aligned with existing Union instruments, including the European Health Data Space, the Pharmaceutical Strategy for Europe and the objectives of

the European Health Union.

Recommendation 3 (on participation, accountability and the rights of patients)

The European Parliament considers that the legislative act to be adopted should strengthen the position of rare disease patients and their families within Union health policy and ensure meaningful participation in governance structures.

The European Parliament considers that the framework should:

– guarantee structured involvement of patient organisations, healthcare professionals and civil society in advisory bodies and evaluation processes at Union level;

– ensure transparency of decision-making, public availability of monitoring data and periodic reporting to the European Parliament and to the Council;

– promote cross-border recognition of expert centres and facilitate patient mobility where appropriate, without undermining national competence in healthcare organisation;

– provide safeguards to ensure that the generation and sharing of health data fully respect fundamental rights, including data protection and privacy;

– promote awareness, training and education initiatives to reduce stigma and improve social inclusion of persons living with rare diseases;

– address the socio-economic impact of rare diseases, including effects on employment, education and gender equality, through coordinated policy approaches at Union level.

The European Parliament considers that individuals affected by rare diseases should benefit from equitable access to timely diagnosis, high-quality treatment and integrated care irrespective of their Member State of residence, and that this objective cannot be sufficiently achieved by Member States acting alone.

Recommendation 4 (on effective implementation, financing and performance convergence)

The European Parliament considers that the legislative act to be adopted should ensure the effective and measurable implementation of the European rare disease action framework through clear operational instruments, financial alignment and enforceable coordination mechanisms.

The European Parliament considers that the framework should:

– establish binding implementation tools, including delegated and implementing acts, to operationalise the European diagnostic coordination mechanism, the genetic diagnosis roadmap, the rare disease research agenda and the Union framework on artificial intelligence in rare diseases;

– ensure that Union financial instruments, including the national regional partnership plan (NRPP), are strategically aligned with the objectives of the Regulation and linked to measurable progress in reducing diagnostic delay, strengthening genomic capacity and

improving equitable access to therapies;

– introduce conditionality mechanisms whereby access to specific Union funding is linked to compliance with agreed screening standards, interoperability requirements and reporting obligations;

– require the Commission to develop a performance scoreboard based on the European rare disease benchmark, enabling objective comparison of Member States and identification of structural gaps;

– provide for systematic data collection and independent evaluation of outcomes, including progress towards the one-year diagnosis objective where medically feasible;

– strengthen cross-border implementation capacity by supporting joint clinical trials, coordinated regulatory pathways for advanced therapies and shared digital infrastructures.
The European Parliament considers that the success of the framework will depend not only on policy design but on enforceable monitoring, transparent reporting and sustained financial commitment, ensuring that convergence in standards and outcomes is achieved across the Union while fully respecting Member States' competences in healthcare organisation.

B. TEXT OF THE PROPOSAL REQUESTED

Regulation (EU) 2026/... of the European Parliament and of the Council on the Rare diseases action framework

of....

THE EUROPEAN PARLIAMENT AND THE COUNCIL OF THE EUROPEAN UNION,

Having regard to the Treaty on the Functioning of the European Union, and in particular Article 114(1) thereof,

Having regard to the European Parliament's request to the European Commission,

Having regard to the proposal from the European Commission,

After transmission of the draft legislative act to the national parliaments,

Having regard to the opinion of the European Economic and Social Committee[1],

Acting in accordance with the ordinary legislative procedure[2],

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1 OJ...
2 OJ...

Whereas:

(1) Although individually rare, they collectively affect an estimated 27 to 36 million people in the Union and therefore constitute a significant public health challenge.

(2) More than 6 000 rare diseases have been identified, the majority of which are genetic in origin, often manifesting in childhood and requiring lifelong, multidisciplinary care.

(3) Approximately 95 % of rare diseases lack an authorised therapeutic option and, where treatments exist, access across Member States remains uneven, resulting in disparities in patient outcomes.

(4) Patients affected by rare diseases frequently experience prolonged diagnostic delays, misdiagnoses and inappropriate treatments, leading to avoidable health deterioration, psychological distress and increased long-term healthcare and social costs.

(5) Rare diseases are characterised by small and geographically dispersed patient populations, limiting the ability of individual Member States to independently generate sufficient clinical evidence, research capacity, specialised expertise and negotiating leverage.

(6) Cross-border pooling of expertise, data and resources generates European added value by overcoming structural scale limitations and enabling efficient research, diagnosis and treatment pathways.

(7) The European Parliamentary Research Service, in its European Added Value Assessment on an EU rare disease action plan of February 2026, concluded that fragmentation of diagnosis, research, data infrastructure and treatment access across Member States results in inefficiencies, inequalities and missed opportunities for improved health and socio-economic outcomes.

(8) The European Economic and Social Committee has in its conclusions and recommendations of 29 November 2024 on the conference for an EU commitment to tackling rare diseases called for a comprehensive European action plan on rare diseases based on solidarity, measurable targets, patient participation and cross-border cooperation, emphasising the social and economic burdens borne by patients and their families.

(9) The Council, in its conclusions of 29 May 2024 on the Future of the European Health Union: A Europe that cares, prepares and protects, has recognised the necessity of reinforced cooperation between Member States to address inequalities in access to healthcare and medicinal products, including in the field of rare diseases.

(10) The establishment of European Reference Networks (ERNs) under Directive 2011/24/EU

of the European Parliament and of the Council[3] has demonstrated the benefits of Union-level cooperation in pooling expertise for rare and complex conditions. However, enhanced governance, sustainable financing, structured integration into national health systems and strengthened referral pathways are required to fully realise their potential.

(11) Significant disparities persist across Member States in the screening programmes of newborns, genomic testing capacity and early detection strategies, resulting in unequal opportunities for timely diagnosis. Voluntary alignment of screening principles and the establishment of benchmark standards can contribute to convergence while respecting national competences.

(12) Earlier and more accurate diagnosis reduces avoidable healthcare expenditure, improves patient outcomes and enhances labour market participation. The establishment of measurable objectives, including a one-year diagnostic target where medically feasible, can guide coordinated action and enhance accountability.

(13) The progressive integration of genomic technologies into healthcare systems requires structured coordination at Union level to promote quality standards, interoperability and equitable access. A Union roadmap can define common objectives and measurable targets without harmonising national healthcare delivery.

(14) Market fragmentation within the Union contributes to disparities in access to orphan medicinal products and advanced therapies, with patients in certain Member States experiencing substantial delays in availability and reimbursement.

(15) Coordinated clinical evidence generation, structured exploitation obligations linked to Union funding, voluntary cooperation in procurement and strengthened collaboration in health technology assessment can enhance equity and efficiency while fully respecting Member States' competence in pricing and reimbursement decisions.

(16) Platform technology master files made available under transparent and non-discriminatory conditions can facilitate the development and scaling-up of advanced therapy medicinal products, particularly for rare diseases with limited commercial incentives, while ensuring high scientific standards and regulatory efficiency.

(17) A structured cross-border exchange framework for hospital exemption advanced therapy medicinal products can facilitate access to treatments addressing unmet needs, particularly for patients who are clinically unfit to travel or affected by ultra-rare conditions, while preserving patient safety, regulatory responsibility and the non-commercial character of the hospital

[3] Directive 2011/24/EU of the European Parliament and of the Council of 9 March 2011 on the application of patients' rights in cross-border healthcare (OJ L 88, 4.4.2011, p. 45, ELI: http://data.europa.eu/eli/dir/2011/24/oj).

exemption framework.

(18) Rare disease research requires critical mass in patient recruitment, data pooling and biobank coordination, which can only be achieved through structured cross-border collaboration and strategic alignment of Union funding instruments.

(19) Artificial intelligence systems have the potential to improve diagnostic accuracy, optimise clinical trial recruitment and enhance research efficiency in rare diseases. A Union-level framework is necessary to ensure that such systems are developed and deployed in accordance with Union legislation on data protection, medical devices and high-risk artificial intelligence, while safeguarding transparency and human oversight.

(20) Fragmented rare disease registry systems and inconsistent coding standards limit the capacity to conduct epidemiological surveillance, generate real-world evidence and support regulatory and health technology assessment processes.

(21) The integration of rare disease data infrastructures into the European Health Data Space established by Regulation (EU) 2025/327 of the European Parliament and of the Council[4] presents an opportunity to enhance interoperability, facilitate secure secondary use of data and support evidence-based policymaking in compliance with Union data protection rules.

(22) The socio-economic impact of rare diseases extends beyond healthcare, affecting education, employment, gender equality and social inclusion, and placing a disproportionate burden on caregivers.

(23) Integrated, multidisciplinary care pathways and cross-border continuity of care are necessary to address the complex and lifelong needs of rare disease patients and their families.

(24) To ensure coherence between early diagnosis, equitable access to treatment, research coordination, artificial intelligence deployment, data interoperability, patient support and governance, a structured European rare disease action framework should be established.

(25) In order to ensure that Union financial support in the field of rare diseases effectively contributes to early detection and reduction of diagnostic delay, access to certain Union funding instruments, including the national regional partnership plan (NRPP), should be linked to the existence or progressive implementation of screening frameworks aligned with Union benchmark standards.

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[4] Regulation (EU) 2025/327 of the European Parliament and of the Council of 11 February 2025 on the European Health Data Space and amending Directive 2011/24/EU and Regulation (EU) 2024/2847 (OJ L, 2025/327, 5.3.2025, ELI: http://data.europa.eu/eli/reg/2025/327/oj).

(26) The establishment of monitoring indicators, a European Union rare disease benchmark and regular reporting obligations is necessary to ensure transparency, performance comparison, identification of structural gaps and continuous improvement across Member States.

(27) A permanent coordination board bringing together Member States, ERNs, patient organisations, researchers and the Commission can strengthen strategic alignment and ensure effective implementation of this Regulation.

(28) Since the objectives of this Regulation — namely reducing fragmentation, enhancing coordination, ensuring interoperable data exchange, facilitating cross-border cooperation in advanced therapies and generating European added value in the field of rare diseases — cannot be sufficiently achieved by the Member States acting alone but can rather, by reason of the cross-border nature and scale of the challenge, be better achieved at Union level, the Union may adopt measures, in accordance with the principle of subsidiarity as set out in Article 5 of the Treaty on European Union.

(29) In accordance with the principle of proportionality, this Regulation does not harmonise the organisation and delivery of healthcare services but establishes coordination mechanisms, targets, reporting obligations and support measures necessary to achieve its objectives.

(30) A Regulation is the appropriate legal instrument to ensure uniform application of coordination standards, data interoperability requirements and governance mechanisms across the Union, thereby preventing further fragmentation that could result from divergent transposition measures. In order to ensure uniform conditions for the implementation of this Regulation, implementing powers should be conferred on the Commission. Those powers should be exercised in accordance with Regulation (EU) No 182/2011 of the European Parliament and of the Council[5].

(31) In order to supplement or amend certain technical non-essential elements of this Regulation, the power to adopt acts in accordance with Article 290 TFEU should be delegated to the Commission in respect of the establishment and coordination of a Union Roadmap for the improvement of genetic diagnosis in rare diseases, the establishment of interoperability criteria and common data standards for the rare disease data infrastructures, the establishment of the structure and functionality of an European rare disease coordination board, and the establishment of an EU rare diseases benchmark. It is of particular importance that the Commission carry out appropriate consultations during its preparatory work, including at expert level, and that those consultations be conducted in accordance with the principles laid down in the Interinstitutional Agreement of 13 April 2016 on Better Law-Making[6]. In

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5 Regulation (EU) No 182/2011 of the European Parliament and of the Council of 16 February 2011 laying down the rules and general principles concerning mechanisms for control by the Member States of the Commission's exercise of implementing powers (OJ L 55, 28.2.2011, p. 13, ELI: http://data.europa.eu/eli/reg/2011/182/oj).

6 OJ L 123, 12.5.2016, p. 1, ELI: http://data.europa.eu/eli/agree_interinstit/2016/512/oj.

particular, to ensure equal participation in the preparation of delegated acts, the European Parliament and the Council receive all documents at the same time as Member States' experts, and their experts systematically have access to meetings of Commission expert groups dealing with the preparation of delegated acts.

(32) The Commission should periodically evaluate the implementation, effectiveness and proportionality of this Regulation, including progress towards diagnostic targets, performance convergence and equitable access to therapies.

CHAPTER I

GENERAL PROVISIONS

Article 1
Subject matter and scope

This Regulation establishes a European rare disease action framework to:

(a) improve early diagnosis and screening;
(b) enhance equitable access to treatment;
(c) strengthen research and innovation;
(d) ensure interoperable data infrastructures;
(e) support integrated care and social inclusion;
(f) reinforce governance and coordination mechanisms.

Article 2
Definitions

For the purposes of this Regulation, the following definitions apply:

(1) ‘rare disease’ means a life-threatening or chronically debilitating condition affecting not more than 5 in 10 000 persons in the Union;

(2) ‘rare disease registry’ means an organised system for collecting uniform data concerning patients with a specific rare disease;

(3)‘ultra-rare condition’ means a condition affecting fewer than 1 in 50 000 persons;

(4)‘patient journey’ means a tool development that captures the natural history of a rare condition and the needs of patients through diary experiences and life-lens of people living with this condition.

CHAPTER II

DIAGNOSIS AND EARLY DETECTION

Article 3
European diagnostic coordination mechanism

1. The Commission shall establish a coordination mechanism to support Member States in reducing average time to diagnosis.

2. The mechanism shall facilitate exchange of best practices, voluntary alignment of the screening principles of newborns, genetic prenatal screening, and development of diagnostic and early diagnostic guidelines.

Article 4
EU diagnostic targets

1. By 1 January 2030, Member States shall ensure that patients with suspected rare diseases receive an accurate diagnosis within one year from the first medical consultation where medically feasible. The Commission shall establish recommendations and guidelines.

2. The Commission shall develop monitoring indicators and milestones in cooperation with Member States.

Article 5
Roadmap for the improvement of genetic diagnosis in rare diseases

1. The Commission shall adopt a delegated act in accordance with Article 18 in order to supplement this Regulation by establishing and coordinating a Union Roadmap for the improvement of genetic diagnosis in rare diseases (the ‘Roadmap’).

2. The Roadmap shall define common objectives, minimum recommended standards and measurable targets for the progressive integration of genomic technologies into national healthcare systems, with a view to reducing diagnostic delay, increasing diagnostic yield and ensuring equitable access across the Union.

3. The Roadmap shall provide for a phased implementation, including:

(a) access to validated next-generation sequencing technologies and accredited genomic laboratories;
(b) interoperable rare diseases registries and secure data infrastructures;
(c) multidisciplinary clinical-genomic evaluation frameworks and defined timelines for diagnosis; and
(d) advanced genomic approaches, systematic data reanalysis and cross-border data

sharing.

4. By 1 January 2030, and biannually thereafter unless the delegated act referred to in paragraph 1 provides for more frequent reporting, Member States shall report on the progress made with regard to the indicators set out in the Roadmap. .

CHAPTER III

ACCESS TO TREATMENT AND AFFORDABILITY

Article 6
Accessibility through fostered development of medicines

Where Union funding under any fund established by the Union contributes in whole or in part to the development of advanced therapy medicinal products intended for the prevention, diagnosis or treatment of rare diseases, beneficiaries must ensure that the exploitation of such results contributes to their affordability, availability, and accessibility in a manner consistent with public health needs in the Member States. The Commission and the Member States may provide for appropriate measures in the grant agreements to facilitate compliance with that requirement. For that purpose, the work programme shall specify additional exploitation obligations that shall include, as a minimum:

(a) the creation of plans for equitable access that outline strategies and timelines for achieving affordability, availability and accessibility for the medicinal product(s);
(b) filing of an application for an open platform technology master file;
(c) accessibility issues related to specific rare diseases.

Article 7
Advanced therapies scaling-up

1. For the purposes of this Regulation, a platform technology master file shall be considered an ‘open platform’ where the owner of the underlying technology makes the relevant regulatory data, documentation and validated scientific components available, under transparent and non-discriminatory conditions, to any third party developing a product sharing the same common regulatory data and scientific components.

2. The Commission shall adopt an implementing act establishing the procedural and technical requirements for the submission, assessment and maintenance of open platform technology master files, with particular regard to their use in the development of advanced therapy medicinal products, including those targeting rare diseases. That implementing act shall be adopted in accordance with the advisory procedure referred to in Article 19.

3. A holder of a hospital exemption authorisation may apply for the establishment or extension of an open platform technology master file on the basis of the investigational medicinal product dossier supporting the authorised product, provided that the platform demonstrates scientific validity, reproducibility and potential applicability to other products based on the same technological principle.

4. The application referred to in paragraph 3 shall be accompanied by a report from the competent national regulatory authority confirming that the platform has been favourably assessed and that its use could facilitate the development, scaling-up or regulatory evaluation of products sharing the same scientific or technological basis, irrespective of whether a marketing authorisation is immediately sought.

CHAPTER IV

RESEARCH AND INNOVATION

Article 8
European rare disease research agenda

1. By...[two years from the date of entry into force of this Regulation], the Commission shall adopt an implementing act establishing a European rare disease research agenda (the 'Agenda'). That implementing act shall be adopted in accordance with the advisory procedure referred to in Article 19.

2. The Agenda shall establish Union-level research priorities in the field of rare diseases, including translational research, advanced therapies, genomic medicine, real-world evidence generation and clinical trial innovation.

3. The Agenda shall ensure strategic alignment and coordination of relevant Union funding instruments and programmes, with a view to maximising synergies, avoiding duplication and accelerating the development of diagnostics and therapies for rare diseases.

4. The Agenda shall promote and facilitate cross-border and multinational clinical trials, including adaptive and platform trial designs, and shall support the integration of research infrastructures, regional biotech hubs, data-sharing frameworks and patient registries across the Union.

5. The Commission shall report periodically on the implementation of the Agenda and its impact on research capacity, innovation output and patient access to novel diagnostics and treatments.

Article 9

Artificial intelligence in rare diseases

1. The Commission shall adopt an implementing act establishing a Union framework for the responsible deployment of artificial intelligence systems in the field of rare diseases, with the objective of improving early diagnosis, clinical decision-making, research efficiency and patient outcomes. That implementing act shall be adopted in accordance with the advisory procedure referred to in Article 19.

2. The framework shall define common requirements for the development, validation and use of artificial intelligence tools applied to rare disease screening, diagnosis, prognosis, treatment optimisation and research, ensuring scientific robustness, transparency, traceability and human oversight.

3. Artificial intelligence systems deployed pursuant to this Article shall comply with applicable Union legislation on data protection, medical devices, digital health and high-risk artificial intelligence systems, and shall ensure interoperability with national and Union health data infrastructures.

4. Member States shall promote the integration of validated artificial intelligence tools into rare disease reference centres, genomic diagnostic pathways and research networks, including through cross-border data collaboration where appropriate.

5. Union financial support for rare disease actions may prioritise projects incorporating artificial intelligence solutions that demonstrably reduce diagnostic delay, improve diagnostic yield or enhance the efficiency of clinical trials and real-world evidence generation.

6. The Commission shall monitor the implementation and impact of artificial intelligence systems in the field of rare diseases and shall report periodically on their contribution to equity of access, quality of care and innovation across the Union.

CHAPTER V

DATA AND DIGITAL INFRASTRUCTURE

Article 10
Interoperable rare disease data network

1. The Commission shall adopt an implementing act establishing a federated European rare disease data network , unifying existing rare diseases data networks and according to the criteria established under the European Health Data Space. That implementing act shall be adopted in accordance with the advisory procedure referred to in Article 19.

2. Registries shall apply common data standards and coding systems. The Commission shall

update and maintain common data standards according to technological improvement, through the establishment of common mandatory criteria, enabling communication and transmission of rare diseases related health data across Europe under criteria of interoperability.

Article 11

Integration with the European Health Data Space

1. Rare disease data infrastructures shall be technically aligned with the European Health Data Space to enable secure secondary use. The Commission shall adopt a delegated act in accordance with Article 18 in order to supplement this Regulation by establishing interoperability criteria and common data standards.

2. The Commission shall ensure the integration of the ERNs as agents of the development of cross-country standards in exchange of cross-border data exchange within Europe as promoters of standardised practices in common data elements, and rare disease registry interoperability.

3. The Commission shall support improved referral pathways between national systems and ERNs.

Article 12

Real-world evidence

1. The Commission shall promote the use of rare disease registry-based real-world evidence to support regulatory and health technology assessment in decision-making of the Member States on rare diseases.

2. Facilitate cross-border data sharing and collaboration among Member States, ensuring compliance with data protection laws and promoting the use of real-world evidence in joint clinical assessments under Regulation (EU) 2021/2282 of the European Parliament and of the Council[7].

CHAPTER VI

PATIENT SUPPORT AND SOCIAL INCLUSION

[7] Regulation (EU) 2021/2282 of the European Parliament and of the Council of 15 December 2021 on health technology assessment and amending Directive 2011/24/EU (OJ L 458, 22.12.2021, p. 1, ELI: http://data.europa.eu/eli/reg/2021/2282/oj).

Article 13

Integrated care pathways

1. Member States shall be supported in developing multidisciplinary care pathways, including psychosocial and mental health support.

2. The Commission shall establish a plan to coordinate the elaboration, publication and update of guidelines on patient journeys according to the state of the art, with ERN's and patient organisations advice and participation. Paediatric patient journeys shall take priority in the design of the implementation of those guidelines.

Article 14

Cross-border continuity of care

1. Cross-border exchange of hospital exemption medicinal products may be authorised when patient mobility is not feasible due to medical condition, treatment duration, or when the expected patient population in the receiving Member State so requires.

2. The receiving hospital may apply for a hospital exemption authorisation in its own Member State, provided that it demonstrates its capacity to replicate the treatment in accordance with the same quality, safety, and process standards as those applied in the originating Member State.

3. In the circumstances referred to in paragraph 1, the competent national regulatory authority of the receiving Member State may allow the cross-border transfer of the hospital exemption-advanced therapy medicinal product under the hospital exemption framework and designate a treating reference centre in the receiving Member State, subject to the authorisation of the competent national regulatory authorities of the originating Member State for the production and export of the product.

4. This shall not require that all manufacturing activities be carried out within a single Member State, provided that the originating site serves as the reference model for quality, manufacturing, and process standards.

5. Manufacturing activities may be outsourced to the originating Member State. Regulatory responsibility shall remain with the medical practitioner and the hospital requesting the hospital exemption in the receiving Member State, in cooperation with its competent national regulatory authority.

6. The implementation of this Article shall be subject to:

(a) the conduct of appropriate comparability studies;
(b) the use of harmonised clinical and quality protocols; and

(c) the capacity to aggregate and analyse clinical data jointly with the data generated by the hospital holding the original hospital exemption authorisation, in compliance with Union data protection rules.

7. Member States shall ensure that the implementation of this Article does not undermine patient safety, data integrity, or the non-commercial nature of the hospital exemption framework.

CHAPTER VII

GOVERNANCE AND MONITORING

Article 15
European rare disease coordination board

1. A permanent European rare disease coordination board (the 'Coordination Board') is hereby established. The Commission shall adopt a delegated act in accordance with Article 18 in order to supplement this Regulation by establishing the structure and functionality of that board.

2. The Coordination Board shall include representatives of Member States, ERNs, patient organisations, researchers and the Commission. The representative of the Commission shall be the Chair of the Coordination Board..

3. The Coordination Board shall establish the priorities in the development of the ERNs, and its joint work programme.

Article 16
EU rare diseases benchmark

1. The Commission shall adopt a delegated act in accordance with Article 18 in order to supplement this Regulation by establishing an EU rare diseases benchmark.

2. The EU rare diseases benchmark shall measure and monitor performance across five strategic pillars:

(a) diagnosis and early detection;
(b) access to treatment and care;
(c) research and innovation;
(d) coordination and governance;
(e) patient-centered outcomes and social support.

Article 17

Eligibility conditions for rare disease funding under the NRPP

1. Access to financial support under the NRPP for strategic projects in the field of rare diseases shall be conditional upon the existence of a national or regional screening programme of newborns and, where applicable, a prenatal genetic screening framework, established in accordance with Union benchmark standards.

2. For the purposes of paragraph 1, Union benchmark standards shall include, as a minimum:

(a) defined screening panels based on scientific evidence, clinical utility, and public health relevance;
(b) nationwide or regionally coordinated coverage ensuring equitable access;
(c) quality assurance and accreditation mechanisms;
(d) effective referral pathways for confirmatory diagnosis and specialised care; and
(e) compliance with Union law on data protection, informed consent, and non-discrimination.

3. Member States applying for support under NRPP shall demonstrate effective implementation or present a binding implementation plan with a defined timeline to achieve compliance with the standards referred to in paragraph 2.

4. The Commission shall assess compliance with this Article prior to the allocation of funds and may suspend, reduce, or condition financial support where the requirements set out herein are not fulfilled.

5. That conditionality shall aim to ensure that Union funding for rare diseases contributes to early detection, reduction of diagnostic delay, and convergence of standards of care across the Union.

CHAPTER VIII

FINAL PROVISIONS

Article 18
Exercise of the delegation

1. The power to adopt delegated acts is conferred on the Commission subject to the conditions laid down in this Article.

2. The power to adopt delegated acts referred to in Articles 5, 11, 15 and 16 shall be conferred on the Commission for an indeterminate period of time from... [date of entry into force of this Regulation].

3. The delegation of power referred to in Articles 5, 11, 15 and 16 may be revoked at any time by the European Parliament or by the Council. A decision to revoke shall put an end to the delegation of the power specified in that decision. It shall take effect the day following the publication of the decision in the *Official Journal of the European Union* or at a later date specified therein. It shall not affect the validity of any delegated acts already in force.

4. Before adopting a delegated act, the Commission shall consult experts designated by each Member State in accordance with the principles laid down in the Interinstitutional Agreement of 13 April 2016 on Better Law-Making.

5. As soon as it adopts a delegated act, the Commission shall notify it simultaneously to the European Parliament and to the Council.

6. A delegated act adopted pursuant to Articles 5, 11, 15 and 16 shall enter into force only if no objection has been expressed either by the European Parliament or by the Council within a period of two months of notification of that act to the European Parliament and the Council or if, before the expiry of that period, the European Parliament and the Council have both informed the Commission that they will not object. That period shall be extended by two months at the initiative of the European Parliament or of the Council.

Article 19
Committee procedure

1. The Commission shall be assisted by a committee. That committee shall be a committee within the meaning of Regulation (EU) No 182/2011.

2. Where reference is made to this paragraph, Article 4 of Regulation (EU) No 182/2011 shall apply.

Article 20
Financial support

1. Support for rare diseases under the NRPP in the post-2027 Multiannual Financial Framework (2028-2034).

2. For the duration of the post-2027 Multiannual Financial Framework, strategic projects in the field of rare diseases may be supported under the NRPP. Such support shall aim to strengthen early diagnosis, equitable access to treatment, research capacity, digital health infrastructure, and cross-border cooperation, with particular attention to reducing disparities between and within Member States.

3. Funding under the NRPP may complement and create synergies with Union programmes, provided that such support remains consistent with the objectives and legal bases of those

programmes.

4. Priority shall be given to projects delivering measurable Union added value, including the development of interoperable rare disease registries, reinforcement of specialised centres and networks, support for clinical research in small populations, and deployment of secure cross-border data systems. Implementation shall be subject to monitoring and evaluation to ensure transparency, effectiveness, and alignment with Union health objectives.

Article 21
Evaluation

1. By ....[2 years after the date of entry into force of this Regulation] and every two years thereafter, the Commission shall submit a report to the European Parliament and to the Council on implementation progress of this Regulation.

2. The Commission shall evaluate this Regulation by...[five years after the date of entry into force of this Regulation].

Article 22
Entry into force

This Regulation shall enter into force on the twentieth day following that of its publication in the *Official Journal of the European Union*.

EXPLANATORY STATEMENT

Rare diseases represent one of the clearest and most compelling cases for structured European action in the field of public health. Although each individual condition affects a small number of persons, the cumulative impact is considerable. Between 27 and 36 million people in the European Union are estimated to live with a rare disease. More than 6 000 such conditions have been identified, most of them genetic, chronic, progressive and frequently life-threatening. For approximately 95 % of these diseases, no authorised treatment exists. The European Parliamentary Research Service (EPRS), in its 2026 European Added Value Assessment on an EU Rare Disease Action Plan, demonstrates that this field is characterised by structural fragmentation, unequal access to expertise and therapies, persistent diagnostic delay, insufficient data interoperability and significant socio-economic burdens on patients and families.

The European Economic and Social Committee (EESC), in its recent opinions and conference conclusions calling for a European action plan on rare diseases, has underlined the urgency of coordinated Union action grounded in solidarity, patient involvement and measurable targets. The Council has similarly acknowledged, in its conclusions on strengthening the European Health Union and on access to medicines for patients in need, that rare diseases constitute a domain where cross-border cooperation and Union coordination are essential to reduce inequalities and improve resilience. Taken together, these institutional positions create a coherent mandate for legislative initiative under Article 225 TFEU.

The present explanatory statement sets out the political, legal and socio-economic rationale for proposing a Regulation establishing a European Rare Disease Action Framework. It explains the design of the plan, the choice of legal instrument and the structured approach across diagnosis, treatment access, research, data infrastructure, patient support and governance.

Rare diseases illustrate the limits of purely national responses. Healthcare organisation and delivery remain within Member State competence under Article 168 TFEU. However, the challenges inherent in rare diseases are intrinsically transnational. Patient populations are small and geographically dispersed. Expertise is concentrated in limited centres of excellence. Clinical trials require multinational recruitment to reach meaningful sample sizes. Data fragmentation prevents robust epidemiological assessment. Market incentives for pharmaceutical development are uneven and depend on Union-level frameworks such as the orphan medicinal product regime. These structural characteristics mean that purely national action inevitably results in duplication, inefficiency and inequity.

The EPRS European Added Value Assessment demonstrates that coordinated European action would generate measurable benefits. It identifies scale efficiencies in research, improved bargaining capacity through coordinated evidence generation, enhanced data quality through interoperable registries and significant reductions in diagnostic delay. The EESC has reinforced this assessment by emphasising that the absence of a comprehensive EU framework leads to avoidable disparities between citizens depending on their Member State of residence. The Council has repeatedly recognised the importance of solidarity mechanisms and reinforced cooperation in access to medicines, including in its conclusions addressing shortages and innovation in pharmaceutical policy.

The phenomenon commonly referred to as the “diagnostic odyssey” illustrates the human cost of fragmentation. Many patients experience years of uncertainty before receiving an accurate diagnosis. During this period, they may undergo inappropriate treatments, suffer psychological distress and experience deterioration that could have been mitigated by earlier intervention. The EPRS analysis highlights the uneven availability of newborn screening programmes and genomic sequencing capacity across Member States. The EESC has called for common principles for early detection and SMART objectives to reduce diagnostic timelines. Council discussions on the strengthening of the European Health Union have equally underlined the importance of early diagnosis and cross-border exchange of expertise. A structured European framework does not harmonise national healthcare systems; it facilitates voluntary alignment, supports common standards and establishes measurable targets that enhance accountability while respecting subsidiarity.

Access to treatment remains deeply unequal across the Union. Although the orphan medicinal product framework has stimulated innovation, disparities in reimbursement, health technology assessment practices and pricing negotiations create delays and divergences. Patients in smaller or lower-income Member States may wait years longer for access to therapies available elsewhere. The EPRS EAVA identifies European added value in joint clinical evidence generation and coordinated approaches that reduce duplication and strengthen Member State negotiating capacity. The EESC has stressed that solidarity and equity must underpin any future action plan, ensuring that no patient is disadvantaged due to geography. Council conclusions on access to medicines for patients in need similarly recognise the necessity of enhanced cooperation to ensure availability and affordability across the Union.

Research and innovation in rare diseases are inherently dependent on cross-border collaboration. The limited number of patients per condition makes national research efforts insufficient. The EPRS assessment demonstrates that pooling biobanks, registries and clinical trial networks increases statistical power and accelerates translational pathways. Horizon Europe funding already supports rare disease research, yet coordination remains fragmented. The EESC has emphasised the importance of patient participation in research governance and the need for coherent long-term strategies. Council conclusions on research and innovation within the Health Union context reinforce the principle that collective European capacity enhances global competitiveness and resilience. A structured action framework aligns existing funding instruments, avoids duplication and creates measurable objectives without imposing harmonisation of national research policies.

Data infrastructure constitutes a foundational pillar of effective rare disease policy. The European Health Data Space represents a transformative opportunity to ensure interoperability and facilitate secondary use of health data. The EPRS EAVA identifies inconsistent coding systems and non-interoperable registries as major barriers to evidence generation and policymaking. ORPHAcodes and common data standards are essential to overcoming these obstacles. The EESC has stressed the need for transparent, ethical and patient-centred data governance. Council discussions on digital health and the EHDS emphasise security, privacy and trust. Integrating rare disease registries into the EHDS enhances both clinical care and research while respecting fundamental rights under Article 16 TFEU.

Rare diseases impose profound socio-economic burdens extending beyond clinical treatment. Families often face reduced employment participation, financial strain and mental health challenges. Care responsibilities disproportionately affect women, reinforcing gender

inequalities. Children with rare diseases require specialised educational and social support. The EPRS assessment quantifies the indirect costs associated with lost productivity and caregiver burden. The EESC has explicitly highlighted the social dimension of rare diseases, calling for integrated care pathways and recognition of caregivers. Council conclusions on social inclusion and disability policy underscore the need to address the functional impact of chronic conditions. An effective action plan must therefore integrate health and social perspectives, ensuring multidisciplinary care and continuity across borders.

Governance is central to sustainability. European Reference Networks represent a major achievement of EU health cooperation, enabling cross-border consultation and pooling of expertise. However, the EPRS analysis identifies challenges related to stable financing, integration into national systems and administrative complexity. The EESC has advocated reinforcing ERNs and embedding them structurally within Member State frameworks. Council conclusions on the European Health Union recognise the need to strengthen cross-border healthcare cooperation. The proposed Regulation therefore provides for a permanent coordination platform, measurable indicators and regular reporting to ensure transparency and accountability.

The inclusion of a chapter addressing Hospital Exemption pathways for advanced therapy medicinal products responds to evolving pharmaceutical legislation discussions. Advanced therapies are particularly relevant for rare diseases. The EPRS EAVA emphasises the benefits that hospital exemption authorisations provide to patients suffering rare diseases and unmet needs. A cross-border exchange mechanism will promote the expansion of best standards of hospital exemption authorisations promoting the uniform application of Union law regarding best scientifical standards of operation. Furthermore, introducing cross-border exchange of HE-ATMPs put patient needs first, particularly of those who are clinically unfit to travel e.g. transplant cases or sufferers of rare conditions. Such exchange should be further strengthening as complementary to the right to cross-border health, and more importantly fully aligned with the scope of the European Reference Networks where expertise travels, not the patients.

The choice of a Regulation as the legal instrument is deliberate and justified. A Regulation ensures uniform applicability across Member States without requiring transposition into national law. Given the cross-border nature of rare diseases, data interoperability and ERN coordination, uniform standards are necessary to avoid further fragmentation. A Directive would risk divergent implementation timelines and interpretations, undermining the very objective of coherence. At the same time, the Regulation is carefully designed to respect subsidiarity. It does not harmonise healthcare delivery or impose binding clinical protocols. Instead, it establishes coordination mechanisms, targets, reporting obligations and frameworks for voluntary cooperation. It relies on incentive measures under Article 168(5) TFEU and internal market coordination under Article 114 TFEU where appropriate. The proportionality principle is respected because the measures address clear cross-border externalities and generate European added value that Member States cannot achieve alone.

The architecture of the plan reflects the six thematic pillars identified in the EPRS European Added Value Assessment. Diagnosis and early detection form the first pillar because timely identification determines long-term outcomes. Access and affordability constitute the second pillar, addressing market fragmentation and inequity. Research and innovation represent the third pillar, ensuring future therapeutic development. Data infrastructure forms the fourth pillar, enabling evidence-based decision-making. Patient support and social inclusion constitute the fifth pillar, recognising the multidimensional impact of rare diseases.

Governance and monitoring provide the sixth pillar, ensuring sustainability and accountability. This design ensures coherence, avoids duplication with existing instruments and aligns with broader EU health strategies.

Financial considerations have been carefully assessed. The EPRS EAVA modelling indicates that reductions in inefficiencies, improved productivity and avoided healthcare costs could yield a positive cost-benefit ratio over a ten-year horizon. The EESC has emphasised that investment in rare diseases is not merely expenditure but a commitment to equity and social cohesion. Council conclusions on sustainable health systems recognise the importance of preventive and coordinated approaches to reduce long-term costs.

Politically, rare diseases embody the principle of European solidarity. Citizens affected by rare conditions often feel invisible within national systems due to the rarity of their condition. European cooperation transforms that rarity into shared strength by pooling knowledge and resources. The action framework aligns with the European Pillar of Social Rights, the Pharmaceutical Strategy for Europe and the objectives of the European Health Union. It operationalises the commitment that access to high-quality healthcare should not depend on geography or disease prevalence.

The Regulation format provides legal certainty, transparency and durability. It anchors the action plan within the Union's legislative framework rather than relying solely on soft coordination. It establishes measurable indicators and reporting obligations that allow Parliament to exercise democratic oversight. It creates predictable structures that facilitate long-term planning for research, data infrastructure and ERN financing. At the same time, it leaves room for Member States to adapt implementation to their healthcare systems.

In conclusion, the European Added Value Assessment demonstrates that fragmented national approaches are insufficient to address the complex, cross-border nature of rare diseases. The EESC and Council have recognised the urgency and legitimacy of coordinated European action. The proposed Regulation establishes a balanced, proportionate and solidarity-based framework that enhances equity, efficiency and innovation while respecting subsidiarity. It transforms existing initiatives into a coherent strategy with measurable objectives and accountability mechanisms. The European Parliament, acting under Article 225 TFEU, therefore calls on the Commission to submit a legislative proposal establishing a European Rare Disease Action Framework that fulfils the Union's commitment to health, dignity and social justice for all citizens affected by rare diseases.

Access to funding for rare disease strategic projects under the National Regional Partnership Plan (NRPP) shall be linked to the existence and effective implementation of appropriate national or regional newborn and prenatal genetic screening plans, aligned with Union benchmark standards.

Member States seeking support under the NRPP for rare disease-related investments shall demonstrate that they have established or are in the process of establishing within a defined timeframe, comprehensive screening strategies that ensure equitable, evidence-based, and ethically governed access to newborn screening and, where appropriate, prenatal genetic screening. Such plans shall comply with Union principles on quality, safety, data protection, informed consent, and non-discrimination.

Eligibility for NRPP funding shall require that screening programmes meet minimum Union

benchmark standards, including defined condition panels based on clinical utility and public health impact; nationwide or regionally coordinated coverage; quality assurance and accreditation mechanisms; integration with specialised centres for confirmatory diagnosis and care pathways; and interoperability of screening data systems with European health data infrastructures.

The Commission shall assess compliance with these benchmark standards prior to the allocation of NRPP resources and may establish corrective action mechanisms where gaps are identified. Priority under the NRPP shall be granted to Member States that demonstrate measurable progress in reducing diagnostic delay through systematic screening and in ensuring equal access across regions.

This conditionality aims to ensure that Union investment in rare diseases is anchored in early detection and prevention strategies, thereby maximising health outcomes, reducing long-term system costs, and promoting convergence in standards of care across the Union.

In addition to screening conditionality, the proposed framework introduces a structured performance convergence mechanism to ensure that Union support translates into measurable improvements in patient outcomes. The Regulation therefore establishes a European Rare Disease Benchmark as a common reference tool for monitoring progress across Member States. This benchmark will include indicators on diagnostic timelines, access to specialised centres, availability of genomic testing, participation in European Reference Networks, registry interoperability and access to authorised therapies. Its purpose is not to rank systems but to identify gaps, guide investment and promote upward convergence.

A central innovation is the establishment of a Commission-coordinated roadmap to achieve, where medically feasible, a maximum one-year timeframe from first specialist consultation to confirmed diagnosis for suspected rare genetic diseases. Persistent multi-year delays remain a major source of avoidable harm, including disease progression, psychological burden and inefficient resource use. By setting a clear Union-level objective with measurable indicators and reporting obligations, the framework introduces accountability while leaving clinical organisation to Member States.

The Regulation also strengthens cross-border diagnostic cooperation through systematic integration of genomic and phenotypic data into interoperable registries aligned with the European Health Data Space architecture. Current fragmentation of coding systems and data standards limits research and health planning. Harmonised classifications and secure cross-border exchange will enable faster identification of ultra-rare conditions and support evidence-based policymaking.

Regarding advanced therapies, the framework supports responsible development and scaling-up of innovative treatments, particularly where commercial pathways are limited due to small populations. By promoting cooperation on hospital exemption pathways and platform-based regulatory approaches, it seeks to ensure high scientific standards, patient safety and legal certainty while facilitating access for unmet needs. Structured exchange between national authorities will reduce divergence and strengthen trust.

Artificial intelligence and advanced computational tools are recognised as key enablers in diagnosis and research. The proposal integrates AI-based diagnostic support systems within the Regulation’s governance architecture. Such systems must comply with Union standards

on safety, transparency, data protection and human oversight. Their deployment aims to shorten diagnostic pathways, assist clinicians in analysing complex datasets and optimise clinical trial recruitment without replacing professional judgment.

Financial coherence is another cornerstone. The Regulation aligns existing Union instruments—research, digital infrastructure and health programmes—under a unified strategic direction. It avoids duplication by establishing a Union research agenda with clear priorities, ensuring that funding contributes to shared objectives such as reducing diagnostic delay, strengthening interoperability and expanding equitable access. This alignment enhances efficiency and sustainability.

Monitoring and evaluation are embedded throughout the framework. Member States will submit periodic implementation reports with quantitative benchmark data. The Commission will publish consolidated progress assessments and may propose corrective measures or targeted support where persistent disparities are identified. This governance model keeps the Regulation dynamic, evidence-based and responsive to scientific developments.

The proposal also recognises patient involvement as a structural principle. Patient representatives will participate in advisory structures related to research priorities, data governance and evaluation of diagnostic pathways. This inclusion strengthens legitimacy and improves policy design.

Overall, the framework is designed to create systemic change rather than isolated projects. By combining measurable targets, financial conditionality, interoperable data systems, regulatory cooperation and structured governance, the Regulation establishes a coherent European architecture for rare diseases, delivering earlier diagnosis, fairer access to treatment and sustained innovation across the Union.

ANNEX: DECLARATION OF INPUT

Pursuant to Article 8 of Annex I to the Rules of Procedure, the rapporteur declares that he included in his report input on matters pertaining to the subject of the file that he received, in the preparation of the draft report, from the following interest representatives falling within the scope of the Interinstitutional Agreement on a mandatory transparency register[1], or from the following representatives of public authorities of third countries, including their diplomatic missions and embassies:

1. Interest representatives falling within the scope of the Interinstitutional Agreement on a mandatory transparency register
Eurordis
Fundació Sant Joan de Déu
SIOP Europe
Brains for Brain (B4B)
Mutualité Solidaris Belgique
AstraZeneca
2. Representatives of public authorities of third countries, including their diplomatic missions and embassies
Spanish Permanent Representation

The list above is drawn up under the exclusive responsibility of the rapporteur.

Where natural persons are identified in the list by their name, by their function or by both, the rapporteur declares that he has submitted to the natural persons concerned the European Parliament's Data Protection Notice No 484 (https://www.europarl.europa.eu/data-protect/index.do), which sets out the conditions applicable to the processing of their personal data and the rights linked to that processing.

[1] Interinstitutional Agreement of 20 May 2021 between the European Parliament, the Council of the European Union and the European Commission on a mandatory transparency register (OJ L 207, 11.6.2021, p. 1, ELI: http://data.europa.eu/eli/agree_interinstit/2021/611/oj).