

A EUROPEAN BLUEPRINT FOR RARE DISEASES

Towards an EU Action
Plan for Rare Diseases

Co-developed with
Europe's rare disease
community



October 2026

EXECUTIVE SUMMARY

WHY EUROPE NEEDS AN EU ACTION PLAN ON RARE DISEASES, AND WHY NOW

For decades, rare diseases have shown what Europe can achieve when it works together. The EU has built strong foundations, including the orphan medicinal products framework, the European Reference Networks (ERNs), Orphanet, the European Rare Disease Research Alliance (ERDERA) and the JARDIN Joint Action. These initiatives reflect a shared understanding that no Member State can meet the needs of people living with rare diseases on its own.

The challenge now is to make the most of what Europe has already built

Existing initiatives have transformed the rare disease landscape, but they have often developed through separate policies, programmes and funding cycles. Their full potential will only be realised if they are better connected, sustained and translated more consistently into national health and social systems.

An EU Action Plan would provide the framework and ambition to do this. It would bring existing efforts together around shared goals, clear responsibilities, dedicated resources and common measures of progress. Rather than creating parallel structures, it would help Europe optimise and sustain what already exists, fill remaining gaps, and turn European cooperation into better outcomes for people living with rare diseases.

The moment is now:

- **The next budget is being decided.** The 2028–2034 Multiannual Financial Framework (MFF) will determine whether Europe can sustain and build on its rare disease investments.
- **Legislation alone cannot connect the whole system.** The Biotech Act, the new pharmaceutical legislation, the Health Technology Assessment (HTA) Regulation, the European Health Data Space, and the Critical Medicines Act can strengthen innovation and access, but cannot alone address diagnosis, data, specialised care, social inclusion, research and treatment access.
- **The mandate exists.** Since the 2021 multi-stakeholder foresight study Rare2030, an Action Plan has been backed not only by patients, clinicians, researchers and industry but also by the European Parliament, the European Economic and Social Committee (EESC) and a large majority of Member States.
- **The world is moving.** The 2025 World Health Assembly Resolution requested the World Health Organization to develop a Global Action Plan, giving the EU an opportunity to lead by example.

THE PROBLEM: 30 MILLION PEOPLE STILL FACE UNEQUAL, FRAGMENTED CARE

For the 30 million people living with a rare disease in Europe, two thirds of whom experience onset in childhood, the journey remains too long and depends on where they live. Gaps persist at every step, for example:

Along the patient journey	What people face today
Diagnosis	Nearly 5 years on average from first symptom to diagnosis; 60% are first misdiagnosed; 40% are never referred to a Centre of Expertise
Data	Rare diseases remain insufficiently visible in health systems, with ORPHAcode use below 20% in some countries
Treatment	Around 95% of rare diseases have no approved therapy; fewer than half of innovative medicines reach patients across Europe
Access delays	578 days on average from EU authorisation to reimbursement decision, rising to more than 900 days in some Member States
Daily life	Eight in 10 live with a disability; 53% struggle to obtain public support; seven in 10 patients and carers reduce or stop work
Economic burden	Unemployment is 23% compared with an EU average of 6.1% ; estimated costs might reach €121,900 per person per year

WHAT THE BLUEPRINT PROPOSES

The Blueprint was co-created over ten months by patients, clinicians, researchers, public authorities, and industry, through engagement and consultations with a Core Group of 36 experts, a survey shared with the European Regional Taskforce that received 236 responses from 33 countries, and a workshop of more than 150 participants in Prague at the European Conference on Rare Diseases and Orphan Products (ECRD) in June 2026.

It proposes the core building blocks of an effective Action Plan:

- **Vision and goals:** a shared vision of “better lives for people living with rare diseases in Europe”, supported by three strategic goals: health and quality of life; equality and inclusion; resilient systems, innovation and competitiveness, alongside SMART targets.
- **Governance:** an Advisory Board bringing together the European Commission, Member States, patient representatives and other stakeholders from the rare disease community.
- **Funding:** better alignment of existing EU and national funding, complemented by adequate dedicated resources and visible rare disease objectives in future EU programmes.
- **Monitoring:** shared indicators, public reporting and independent evaluation.
- **Patient partnership:** embedded throughout governance, implementation and evaluation.

18 PRIORITY ACTIONS FOR EU-LEVEL IMPACT

The Blueprint identifies 18 priority actions across six areas where EU coordination can make the greatest difference.

Thematic area	Priority actions
Earlier, faster and more accurate diagnosis	• Structured diagnosis and referral pathways • Coordinated European guidance on newborn screening • Improved access to genetic testing and genomic sequencing
Specialised and highly specialised healthcare	• Updated national rare disease plans • A sustainable and better integrated ERN framework • Improved cross-border access to ultra-rare and highly specialised care
Integrated care and social inclusion	• Person-centred integrated care pathways • Better access to social services and protection • Formal mechanisms for patient and family co-design
Optimising the data ecosystem	• Systematic use of ORPHAcodes • Stronger registries and data linkage • Better integration of rare disease data into the European Health Data Space
Innovative and needs-led research	• Better aligned EU investment • Meaningful patient partnership in research • Connected and sustainable research infrastructures
Available, accessible and affordable medicines and medical devices	• Better aligned evidence generation and assessment • Stronger compassionate-use frameworks • EU solidarity and coordinated access mechanisms

WHY IT MUST BE DONE AT EU LEVEL

Around 85% of rare diseases affect fewer than 500 people in the EU, so no single country can create the scale needed for shared data, research infrastructure, cross-border care or highly specialised expertise. EU action can strengthen national plans through common objectives and indicators, while respecting national responsibilities. It can also help connect Europe's existing rare disease assets more effectively. In doing so, equity and competitiveness can reinforce each other: better-connected data, trial-ready networks and pooled expertise can improve access and research efficiency while making Europe more attractive for life-science investment.

OUR ASK

We call on the European Commission to propose an EU Action Plan on Rare Diseases, using this Blueprint as the community's contribution to its design. The Plan should include:

- A shared vision, common goals and SMART targets linking EU and national action.
- Dedicated resources under the EU's Multiannual Financial Framework (MFF) 2028–2034.
- Patient partnership embedded in governance through a multi-stakeholder Advisory Board.
- A common monitoring framework with public reporting and independent evaluation.
- Delivery of the Blueprint's 18 priority actions.

We call on the Council and Member States to support this proposal, including through a renewed Council Recommendation on rare diseases by the end of 2027. Building on Europe's existing foundations, an EU Action Plan would close gaps across the patient journey, strengthen innovation and help translate European science and expertise into better outcomes for people living with rare diseases across all health systems.

"At the Centre of the Circle" - Turkey





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