

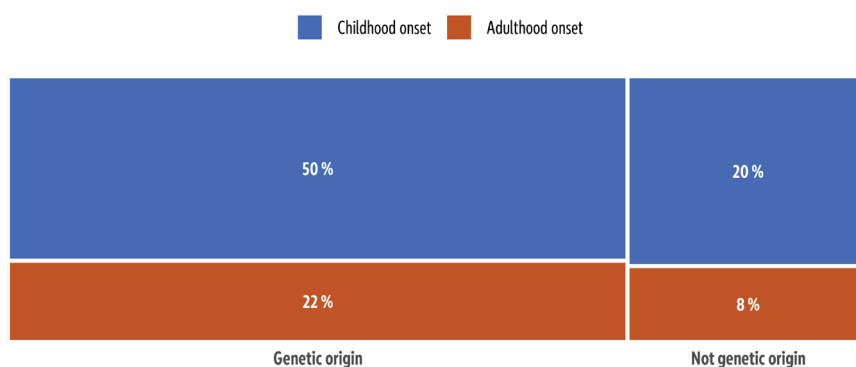
EU rare disease action plan – Main findings from the European Added Value Assessment

Stronger, coordinated EU action to address the diagnosis and treatment of rare diseases could offer value for patients, their families, healthcare providers, researchers, and pharmaceutical developers and society. Drawing on a review of the scientific literature, interviews and case studies, [this study](#), produced at the request of the European Parliament's Committee on Public Health, identifies 31 possible measures for an EU rare disease action plan and examines the drivers of their potential EU added value.

What is the problem?

An estimated 27-36 million people in the EU have a rare disease. Rare diseases for the large part affect children and have genetic origins (see Figure 1). Policy and medical approaches to addressing rare diseases vary across Member States – from development of treatments to screening and sharing of information between healthcare providers and expertise centres – and generate costs for society. These costs are multidimensional and are reflected in sustained healthcare utilisation over the life course, productivity losses, reduced labour market participation among patients and caregivers, and increased reliance on informal care.

Figure 1 – Categorisation of rare diseases



Source: EPRS, based on [EURORDIS](#) and produced by Marco Scipioni.

Which EU measures could be particularly beneficial and why?

The study identified 31 measures across six thematic areas that have clear potential to reduce the health, social and economic burden associated with rare diseases and could be suitable for inclusion in an EU rare disease action plan (see Figure 2). European Reference Networks (ERNs) were found to be a key driver of EU added value across the thematic areas. Findings for each of the six thematic areas are presented below.

Theme #1: Diagnosis and screening

Prolonged diagnostic delays remain a defining feature of rare disease pathways. Further EU action in the form of coordination, guidance, benchmarking and knowledge-sharing could generate EU added value; more specifically, added value could be generated via expert coordination on newborn screening, stronger links between [Orphanet](#), ERNs and primary care, as well as expanded use of telemedicine. Such measures could help to reduce fragmentation across Member States, accelerate learning and improve equity, while respecting national competences.

Theme #2: Access and affordability of therapies

Market authorisation, granted at EU level by the European Medicines Agency, does not equate to actual patient access, as pricing and reimbursement decisions remain a national competence. Moreover, there are persistent market failures driven by small patient populations, high development costs and fragmented national pricing and reimbursement decisions. EU action to help pool demand for new treatments, expertise



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and evidence through coordinated post-marketing evidence generation, voluntary solidarity-based mechanisms, closer alignment between regulatory and HTA processes, and targeted support for ultra-rare conditions, could reduce duplication, strengthen negotiating capacity and support more timely and equitable access.

Theme #3: Research and innovation

In the current framework, it is unlikely that Member States could independently generate the critical mass of patients, data and expertise required to address thousands of rare diseases due to their low prevalence. While the EU has already delivered benefits through initiatives such as the European Rare Disease Research Alliance (ERDERA), gaps remain. Further EU action to pool resources, align research agendas, support shared infrastructures and embed patient involvement could help in reducing duplication and improving efficiency and impact, especially in advanced and emerging approaches such as gene therapies, cell therapies (e.g. CAR T-cell), and next-generation sequencing-based diagnostics.

Theme #4: Data sharing and registries

Rare disease data remains scarce and highly fragmented, limiting their value for research, care and policymaking. Although initiatives such as [Orphanet](#), [European Rare Disease Registry Infrastructure](#) (ERDRI) and ERN registries provide important foundations, uneven standards and governance continue to impede interoperability. Coordinated action to promote common standards, systematic use of [ORPHAcodes](#), federated infrastructures and integration with the European Health Data Space are expected to help internalise cross-border externalities, reduce duplication and unlock long-term benefits.

Theme #5: Patient and family support, social inclusion and care







There is wide variation across Member States with regard to disability recognition, psychosocial support and integrated care. EU added value was identified in measures fostering convergence through guidance, shared frameworks and capacity-building, supporting integrated and person-centred care pathways, functioning-based disability recognition and EU-wide evidence generation. Such measures could help to reduce inequalities including gender inequalities, as women disproportionately assume informal caregiving responsibilities, and indirect economic costs.

Theme #6: Governance and cross-border coordination

The impact of ERNs remains constrained by uneven integration, coverage and sustainability. Further EU action could be taken to strengthen governance, national integration, geographical coverage and horizontal coordination at EU level, including clearer referral pathways, improved reimbursement arrangements for virtual cross-

border consultations, and exploration of stable financing and legal status mechanisms. It could also help to fully realise their added value and to ensure more coherent and efficient rare disease policy implementation across the Union.

Figure 2 - Areas for EU action and potential EU added value

Theme	Potential EU added value
 Diagnosis and screening	>> Shorter diagnostic pathways; earlier detection and treatment; reduced avoidable disease progression, psychological distress and downstream healthcare costs
 Access to therapies	>> Reduced inequalities in access; improved predictability for patients and payers; lower inefficiencies from fragmented national decision-making
 Research and innovation	>> Faster translation of scientific advances; more efficient use of public investment; accelerated development of diagnostics and therapies for unmet needs
 Data sharing and registries	>> Reduced duplication and transaction costs; stronger evidence for clinical, regulatory and policy decisions; improved system efficiency
 Patient and family support	>> Reduced socio-economic burden on patients and carers; improved continuity of care; mitigated impacts on employment, education and informal care
 Governance and cross-border coordination	>> Reduced inefficiencies from fragmented expertise; better use of EU instruments; more equitable access to specialised care across Member States

Source: European added value assessment, EPRS.

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