



POLICY BRIEF – FOR A MATURE EUROPEAN REFERENCE NETWORK SYSTEM IN 2030

A summary of the EURORDIS Recommendations to achieve a Mature ERN System by 2030
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“WE BELIEVE THE ERNS ARE THE SOLUTION THAT WILL PROVIDE A BETTER FUTURE FOR THE NEXT GENERATION”

We, the Rare 2030 young citizens, represent the next generation of patients, advocates and leaders in the rare disease community, safeguarding our future.

Our strength comes from our diversity: the different countries that we come from and from the experiences we draw upon – as patients, students, young parents and young doctors. Whilst we come from diverse backgrounds and hold different, complimentary experiences, **we share the same ambitions and hope, and are united in our vision of our future.**

Many people living with a rare disease advocate, not for themselves, but for the generation yet to come, who will also suffer from the devastating impact living with a rare disease. **We call on the leaders of today, to listen to the voices of the leaders of tomorrow, so we inherit the solution and not the problem.**

We believe the European Reference Networks are the solution that will, when fully realised, provide a better future for the next generation: alleviating the suffering from rare diseases and offering our community a future, when many of our generation have already lost the fight to their rare disease. We choose life, we choose to live.

We call today on policy makers, at national and European level, to commit to take forward the vision presented in this paper for us, and with us, to ensure it becomes a reality.

As the leaders of today you can make a meaningful difference, to drive change for our generation, and the generations of rare disease patients, carers, doctors and policy makers to come.

European Reference Networks offer a once in a lifetime opportunity. Let us not let this opportunity slip from our fingers! The ERNs have grown as we have grown to become networks that truly deliver for people living with rare diseases and complex conditions. **We cannot lose pace now.”**

The Rare 2030 Young Citizens



OUR VISION IS FOR A MATURE EUROPEAN REFERENCE NETWORK (ERN) SYSTEM THAT LEAVES NO PERSON LIVING WITH A RARE DISEASE IN UNCERTAINTY REGARDING THEIR DIAGNOSIS, CARE AND TREATMENT.

ERNs promise to share...

Knowledge sharing at scale will only succeed in an inclusive ecosystem, where ERNs coordinate with the national healthcare systems and are fully integrated with local services. **We expect to see real collaboration between the right experts**, supported by effective tools and **we want the ERNs to be the first-stop, go-to place for trusted information on all rare diseases.**

ERNs promise to care...

Member States should ensure that ERNs' services are accessible to any patient at any moment. Patients from any country should get referred to the best European experts to get proper diagnosis and treatment. The signature of care under an ERN, should be care based on the leading knowledge and experience of a multidisciplinary team, empowering patients in their care. **ERNs may contribute to improve our quality of life by providing good standards of care and guidelines, but health authorities, hospital managers and clinicians must ensure that these are implemented at local level** and that people living with a rare disease can access the right care.

ERNs promise to cure...

Our hope is that **ERNs will allow rare disease patients to secure a diagnosis within 6-12 months from coming to medical attention.** At the same time, we expect the ERNs to accelerate the development and uptake of treatment options for rare diseases, as they integrate European-wide clinical research and care settings, supported by registries.

INTRODUCTION

European Reference Networks (ERNs) promise to *Share, Care and Cure* to tackle the public health challenges that the rare disease community face. To deliver on this promise experts must unify within the Networks, and ERNs must be integrated within healthcare systems across the European Union. As the Networks move to provide greater geographical and disease coverage, under the second call for full members and as they evolve to conform a new European health infrastructure, community action is needed at a national level to mature the environment that Networks will need to connect with. The ERN-model should be an extension of national healthcare systems, allowing them to provide an additional dimension of EU-wide networked care that supplements the multi-disciplinary care provided at national level.

Whilst the idea of cross-border healthcare cooperation for complex and rare conditions, powered by European Reference Networks, took a decade to germinate and find its way into EU legislation, the consolidation of this system now finds itself in a fast-moving political environment. Indeed, the need - and willingness - for more European coordination in health is now growing and is at the heart of the Commission's proposal to build a [European Health Union](#). The next stage of the Networks' development should harness this drive for greater cross-border cooperation on healthcare and be guided by a vision of a mature ERN system in 2030 that will propel the Networks from good to great!

EURORDIS-Rare Diseases Europe, and the community of people living with a rare disease, want to continue to help shape the future ERN system to ensure that the Networks realise their full potential to tackle the public health needs of rare diseases, once and for all. This paper is a summary of the [Recommendations to achieve a mature ERN system by 2030](#). This brief starts off with an overview of the patient population needs that the ERN system should aim to address (part 1). It then describes the vision for a mature ERN system in 2030, highlighting in particular what needs to be in place at national level (part 2), how the Networks' structure should evolve (part 3) and a framework for what services and functions ought to be in place in ten years-time (part 4). Our recommendations can also be found at the end of the paper.



EURORDIS would like to sincerely thank the patient and clinical leads, who have been instrumental in the development of the vision for a Mature ERN system, drawing on their involvement in the development of the Networks.

PART 1 RARE DISEASE PATIENT POPULATION HEALTHCARE NEEDS

With rare, comes rarity. Rarity of patients, of experts and of knowledge, with a limited evidence base and lack of available treatments. As many diseases are progressive, degenerative, disabling and life-threatening, these factors have a significant impact on the quality of life and life expectancy of people living with a rare disease, and bring with them a wide spectrum of needs. Whilst each rare disease is unique, there are commonalities that unite the individual, isolated and often invisible cases, to create a critical mass of more than 6,000 rare diseases, affecting 30 million people in Europe. The latest [epidemiological estimates](#) on the prevalence and incidence of rare diseases show a highly skewed population profile. The vast majority of people living with a rare disease (>98%) are affected by one of the 390 most prevalent diseases (more common than 1 per 100,000), whereas the remaining 2% have an extremely rare condition (prevalence less than 1 per 100,000)¹.

Patient population needs

The individual needs of each patient with a rare or complex condition can vary significantly, requiring specialised knowledge and complex healthcare interventions. Healthcare for complex and rare diseases benefits from drawing on the experience of a network of experts, investing their knowledge and experience to enrich the evidence base that can be employed locally to treat the patient.

Patients often report long delays to securing accurate and timely diagnosis, which can increase the complexity and life-threatening morbidities, as well as inflicting a huge psychological burden. In fact, the consequences of living with a rare disease are far reaching as there is often a severe impact on the everyday professional and personal lives of patients and families. These needs increase exponentially for the people affected by extremely rare conditions, requiring greater cross-border healthcare collaboration to meet them. For these people, there are greater health inequalities in accessing care, a lack of adequate services and a limited number of available, effective and accessible treatments.

The role of the ERNs in addressing patients' needs

The European Reference Networks were specifically set up to address the needs of all patients living with a rare disease or complex condition, and not restricted exclusively to the populations served by the individual centres that conform these Networks. In addition, each Network should understand the full impact of the conditions that it covers, identified through a needs assessment study in order to be able to effectively address these needs and monitor the impact of this needs-led approach. The Networks need to be guided by clear and ambitious strategic goals that are driven by and aligned to meet these needs.

PART 2

HEALTHCARE SYSTEMS POSITIONED TO MEET THE NEEDS OF THE RARE DISEASE COMMUNITY

In preparation for the establishment of the European Reference Networks, the [2009 Council Recommendation on an action in the field of rare diseases](#), encouraged Member States to identify “appropriate Centres of Expertise throughout their national territory by the end of 2013 and consider supporting their creation”. Building the EU capabilities in rare diseases was therefore predicated first on Member States, that were called to organise expertise and capacity within their own healthcare systems. Subsequently, the European Commission Expert Group for Rare Diseases (EUCERD) set out specific recommendations to support Member States in organising healthcare systems for rare diseases. A critical component of these recommendations was the selection process for Centres of Expertiseⁱⁱ, envisioned to be the pivotal structure of the ERN system, connecting the wider European network with the national healthcare system.



Variation in the readiness of healthcare systems

The maturing of the national healthcare ecosystem for rare diseases and the implementation of the EUCERD recommendations, still remains to be realised across the EU27. Whilst some national systems now have a selection and accreditation system in place, on which they build the endorsement of centres to join the Networks, other countries’ systems are more informal. The variation in the endorsement process of healthcare providers has created an imbalance in the application process to join the ERNs and has also caused variation in the number of members from different Member States.

The lack of a common, standardised endorsement by Member States of their respective Centres of Expertise to join an ERN has had as a consequence the lack of a clearly defined role and mandate nationally for the majority of the ERNs’ members. **This directly impacts the ability of these healthcare providers to formally act and take the necessary steps that would connect the ERNs into the national healthcare system.**

The EUCERD recommendationsⁱⁱⁱ, irrespective of the maturity of the different national healthcare systems, remain as relevant today as the day they were published. It is worth reminding that the strength, responsiveness and potential of the ERN system is directed linked to:

- i. the quality of the selection and endorsement of centres within their respective healthcare systems; and
- ii. the level of investment and support^{iv} provided by Member States needed for Centres of Expertise to fulfil the EUCERD recommended for the Quality of Centres of Expertise^v. Investment in the Centres of Expertise, will both strengthen national healthcare systems, offering valuable resources to frontline services, as well as the ERN system as a whole.

Shifting the focus to the national level

At this stage, it is now critical to shift some of the political, financial and administrative focus to the national level, and to increase the support to the Expert Centres so that they can contribute to the work of the ERNs, but also to work on their behalf at national level.

Community action, supported by the European Commission, similar to the [EUROPLAN national conferences](#), ideally in the framework of an EU Joint Action for Integration, would be instrumental to stimulate the national debate on how the ERNs can complement and enhance the national healthcare system. As well as defining the role of ERNs' members in their own national healthcare systems in order for them to connect the national health services to the ERNs. Hospital managers and national health authorities should agree on the support that these centres require to enable them to fulfil the mission and mandate set out in the EUCERD Recommendations and the Council Recommendation. In this way, the clinical units that have joined an ERN will be equipped to draw on the capacities and expertise of the whole ERN to improve care for their patients and build the competencies of their national clinical communities, thus enabling local populations to benefit from improved outcomes that are associated with centralisation of knowledge under an ERN^{viii}.



PART 3 STRUCTURE AND SCOPE OF THE ERNS

The original vision for the structure and extension of ERNs was based on a step-wise approach to ensure all rare diseases had a home^{viii}. The Networks were founded on the principle of multi-disciplinarity at a Network level that would allow different medical experts to collaborate and access the Networks' knowledge assets to treat these multi-system rare diseases. Although the ERNs have started to develop inter-ERN working groups for individual multi-system rare diseases, this collaboration must be extended.

Optimal Structure for the ERNs

The 24 ERNs set out the strategic ambition to pool and disseminate knowledge, as well as to provide specialist advice for all rare diseases and complex conditions included within the Networks' scope. However, three important gaps remain - rare gyneco-obstetrics, benign tumours and highly specialised mental health. In addition, the Networks have initially focused their operational activities on around 1000 rare diseases and complex conditions^{ix}. Whilst the Networks have committed to cover all rare and complex conditions, today many diseases are only formally in the Networks without any specific collaborative activities being undertaken to tackle the healthcare needs of these rare diseases.

The inclusion of any given disease in the scope of the Networks should not be an empty promise. Instead, it should provide assurance that this is based on clearly defined criteria for the selection and inclusion of experts for each rare disease or complex condition, as a quality and safety safeguard. Any disease expansion should be sustained with a commitment for concrete collaborative activities under the Network: for example, the development of consensus statements or clinical practice guidelines, specialist advice, research and educational activities. The Networks are encouraged to develop a step-wise roadmap of activities that outlines the specific collaborative activities for each of disease under their strategic (formal) scope, including revisions of existing and development of new specific criteria. In addition, adequate funding should be made available to match the Networks' ambitions captured in their roadmaps and the EU commitment to tackle all rare diseases and complex conditions, leaving no one behind.

Achieving Equitable Coverage

The Networks are moving towards greater geographical coverage, increasing the number of countries represented in each Network by a full member or an Affiliated Partner^x that increased in 2019 from 44% to 74% coverage and from 3 to 10 Member States with at least one healthcare provider in all 24 Networks^{xi}. Further expansion in terms of geographic coverage is expected when the new full members join the ERNs in 2021.

However, the existing forms of affiliation will not ensure enough inclusivity. Whilst a full member might hold expertise in one or several conditions that fall under the scope of a single Network, it will very seldom have the expertise to cover all the conditions grouped under that ERN. However, the current rules prevent Member States to designate Associated Centres to join a Network if there is already a full member from that country participating in the ERN. Member States should be able to designate Associated Centres when a country has a full member in a given Network, as long as these centres bring in complementary expertise to that of the full members established in their country.

Investing to establish a sustainable system

The Networks' size and scope should be defined based on the specific needs of each thematic grouping of rare diseases and complex conditions, and driven by patient-professional partnership.

The Networks are heterogeneous in size and scope, and the current funding model based on a standard block grant fails to respond to the significant variation of the Networks' structures. The Court of Auditors also highlighted last year the difficulties around sustainable funding and called on the Commission to define a sustainable and long-term funding framework for the ERNs^{xii} by 2022.

The public funds to sustain the ERNs' operations should have a direct link and be proportionate to their size and scale of ambition and activities, through a 'cost and volume' model, that would combine a fixed payment for all Networks of the same amount regardless of size (to cover core structural costs, including administrative and management support, translation, etc). This funding should be complemented with an additional payment proportionate to the size of the Networks (to cover networking activities and logistics).

Additionally, ERNs lack the policies and mechanisms to channel funding from other sources (industry, private donations, CSR, etc). The long-term funding framework for the ERNs should take into consideration all possible sources of funding and define any central functionalities and policies to support ERNs' financial management and governance, including adequate support to enable meaningful patient engagement.

Valuing Patient Involvement

Patient representatives' involvement within the Networks is growing in strength and value. However, there is significant variation in how meaningful their involvement is. Patient representatives work on a voluntary basis for the Networks, are not compensated for their work nor the time invested in ERN activities, and the large majority of the ERNs lack the resources to support patient engagement. Patient involvement in the Networks must be fully legitimised and supported by clear and transparent rules for patient engagement, adequately support and fairly compensated.

International Partnerships

ERN legislation requires that the Networks forge international partnerships and collaboration. However, this has not yet been operationalised. The European Commission, Member States and the ERNs should explore with the WHO European Office the possibility of establishing collaborations with the 59 countries in the WHO European region to pool knowledge, data and efforts to tackle the public health needs of complex and rare conditions and explore collaboration with other international clinical networks.

The hallmark of rare diseases and complex conditions is that expertise and knowledge are scarce, evidence is limited, and care is often fragmented. Together these factors diminish healthcare services' capacity to respond to the complex, multi-system needs associated with these diseases, and result in poor access to adequate care for many people. The information and knowledge sharing linked to the activities developed by the Networks turn each ERN into a learning system, each with an emphasis on different areas (diagnosis, research, treatment options, surgical procedures, therapies, quality of life, etc.) depending on the characteristics of the conditions that fall within their scope. A mature ERN system should be made up of networks that are fully equipped to perform collaborative activities on care, knowledge sharing, training and research as described in this section.

4.1 Care

ERNs are first and foremost about healthcare. The Networks' *raison d'être* is to improve access to diagnosis, treatment and to provide high-quality healthcare for patients who have a condition that requires a particular concentration of resources or expertise^{xiii}. This central philosophy is encapsulated in the Networks signature – Share, Care and Cure. The rare disease community expects the ERNs to provide a home for all rare diseases, where experts are recognised, visible and accessible, and where expertise travels, not the patient.

For the ERNs to improve access to high-quality care, there needs to be:

- a secure and efficient system to manage, share and store health data;
- clearly defined cross-border referral and patient pathways that connect each Network with the EU27 healthcare systems and;
- the provision of specialist advice under a robust clinical governance that assures the public of its quality.

This infrastructure is still under development. The Clinical Patient Management System (CPMS) provides the platform for the Networks' experts to connect and conduct virtual panels. However, the referral systems from local services have not yet been defined. The Networks need to extend their reach, beyond the +1000 cases that have been reviewed on the CPMS system, to connect with local services and support the rare disease community to secure specialist advice under the Networks.

The ERNs governance framework needs to be water-tight to safeguard quality and patient safety. For this to happen, the Networks' specific criteria, must be continuously reviewed, to ensure a similar level of granularity and equal validation across all conditions. In addition, Network members should be able to demonstrate on an ongoing basis that they uphold the required level of competencies.

The ERN model of cross-border healthcare needs to be rolled out across all Member States, through:

- the agreement and implementation of a common referral pathway;
- clearly designated hospitals as referral management centres into the ERNs;
- the development of a suite of digital tools and services; and
- a clear and transparent fair-pricing and reimbursement model for cross-border virtual care.

4.2 Knowledge Sharing

The initial phase of the Networks' development has been focused on forming partnership of experts from across the EU and building a foundation of trust between them and also with the patient representatives. Trust is a fundamental element to establish an effective collaboration^{xiv}. Collaboration and learning together lead to successful, effective networks^{xv}, while collaborative working and knowledge sharing has also shown to improve healthcare outcomes^{xvi}.

The Networks have not yet fully drawn on their true potential in sharing their experiences and expertise in a structured way within and outside each Network. There remains the need for a common methodology to support the development of both clinical guidelines (and clinical decision support tools) as well as clinical pathways. ERNs need a common methodology that goes beyond the value of GRADE methodology^{xvii} and is adapted to the specificities of rare and complex conditions. A methodology that balances the weight of both published evidence and the wealth of experts' experience and knowledge within the Networks. To support the Networks to generate new evidence, best practice and knowledge, additional direct funding is urgently needed for the development of new guidelines and consensus statements.

In addition, the Networks need to continue developing their registries and collect clinical and other outcome measures that can be used to support experts to identify emerging best practice.

The Commission's Assessment, Monitoring, Evaluation and Quality Improvement System (AMEQUIS) will be critical to shape the ERN system as a continuous quality improvement and learning system. However, further resources are needed to curate and safeguard the quality and integrity of the ERNs' data and knowledge.

Further action is also required to determine how the knowledge assets from the ERNs are disseminated and implemented locally, thereby supporting the development of competencies in national healthcare systems. The Board of Member States, ERN members and ERN hospital managers should agree on how to facilitate the uptake and implementation of these knowledge assets at local level.

4.3 Training & Education

Facilitating the mobility of expertise and delivering training within and outside the Networks are two of the core objectives of the ERNs^{xviii}. The Commission and the Networks have begun to take concrete action to enhance and share their knowledge and expertise outside the Networks, through the Expert Educational Webinars Programmes, as well as within the Networks through the Short-term Mobility and Exchanges of Healthcare Professionals. However, the ambition (and potential) for the Networks to promote and deliver medical training and education activities in all Member States remains yet to

be fulfilled. Significant increased support, resources and e-training tools are needed to unlock this potential and support the development of local healthcare systems competencies in rare diseases.

The Networks should expand their educational and teaching activities, jointly with Member States, professional societies, rare disease patient communities and medical training institutes to support the development of local healthcare systems competencies in rare and complex conditions. This is a crucial step to develop the next generation of experts, specifically to raise awareness of rare diseases and their red-flag symptoms among medical students. In addition, there should be a mutual exchange of knowledge between full members and Affiliated Partners.

The European Commission should provide additional funding to develop innovative training modules and interactive tools for the ERNs. It should aim to establish an integrated educational strategy for the development of cross-border supra-specialised training.

4.4 Research

For people living with a rare disease, clinical research is perceived as a natural extension of healthcare, as the majority of these diseases lack a cure. Clinical Research Networks embedded in the ERNs will allow clinicians and scientists in their multi-disciplinary teams, as well as patient representatives, to:

- work together to advance high quality clinical research on rare diseases;
- ensure compliance with regulatory and Health Technology Assessment standards;
- facilitate the systematic collection of outcome measures that are useful for clinicians and relevant for patients and simplify study enrolment and data sharing.

The expectation is that ERNs will facilitate the translation of new therapies and new approaches, such as repurposing, into innovations in routine clinical practice. However, to realise this potential, the Networks need first to consolidate their research capacities and establish partnerships with national and international rare disease clinical research networks as well as with the wider rare disease research ecosystem.

To support these efforts, the 24 ERNs will work together, under a Support and Coordinated Action funded by the Horizon 2020 programme, to deliver in 2025 the foundations of a common research infrastructure that will provide research-related services to all ERNs covering four domains: clinical research, data management, engagement and dissemination, and administrative support. In parallel, each ERN should agree to conduct at least five new natural history studies over the next two years, which will provide insight into the causes and progression of the diseases, as a way to prepare for clinical testing and improve their chances to secure funding to develop clinical trials.

Significant additional funding from different sources, and a fit-for-purpose funding governance model, will be required to fully deploy this common research infrastructure and its services as well as to deliver collaborative clinical trials.

CONCLUSION

The time is right, for the European Reference Network community to seize the dynamic, fast-moving political and policy environment. The time is right to take affirmative action in the development of the Networks' system, to ensure it is bold and far-reaching. The needs of the rare disease community demand decisive action now, to propel the Networks into a powerful, impactful model that the rare disease community deserves. The vision of a Mature ERN system sets out the direction and the destination we, as a community, need to reach, if we are to succeed in turning the tide of suffering and shortened lives many of us face. We cannot let the opportunity ERNs offer us to be missed or fall short of its mark.

We must praise the dedication, solidarity and drive shown by the clinicians and patient representatives that have been involved in the Networks in their formative years.

However, we now call on all of the EU institutions and its Member States, to deliver on the ERN's promise *to share, to care and to cure*, for all not just the few. We must now grasp the opportunity that the ERNs offer, draw on the commitment from the European Union to invest in the ERN system and make it a permanent structure and make a gear change in order to move the system from good to great!





WHAT NEEDS TO HAPPEN TO FULFIL THE VISION OF A MATURE ERN SYSTEM BY 2030

These are our recommendations to deliver on the ERNs promise to share, care, cure. Further details on each of these recommendations can be found in our [full vision paper](#).

PART 1 RARE DISEASE PATIENT POPULATION HEALTHCARE NEEDS

- The European Commission should fund a needs assessment study** to capture the current needs and expectations of the rare disease population and map the ERNs evaluation framework against the rare disease population needs. Target deadline
2021

PART 2 HEALTHCARE SYSTEMS POSITIONED TO MEET THE NEEDS OF THE RARE DISEASE COMMUNITY

- Member States should define common guiding core principles for the endorsement of their national centres of expertise to participate as HCPs members in the ERNs.** Target deadline
2022
- Member States must define a process to identify and designate national Centres of Expertise** where there is none, with a quality improvement approach, ultimately to progress towards a model based on benchmarked health outcomes of clinical services, integrating the opinion and needs of the patient community. Target deadline
2025
- The European Commission could advance the integration of ERNs into national health systems,** by funding national multi-stakeholder workshops to facilitate discussions and actions on integration into each of the EU27 national health systems. Target deadline
2021
- Member States should invest to strengthen the capacities of Centres of Expertise** so they can fulfil the vision set out in the EUCERD Recommendations on Quality of Centres of Expertise. Target deadline
2025
- Member States should enable and strengthen networking at national level by establishing national rare disease reference networks,** updating national RD National Plans or Strategies and engaging with clinicians and patients to set them up and drive their implementation. Target deadline
2025
- Member States and ERNs should collaborate to develop referral pathways to the ERNs and care pathways** that support the smooth transition of affected individuals to adult services and to the ERNs, providing continuity of care. Target deadline
2022

PART 3 STRUCTURE AND SCOPE OF THE ERNS

- 1** Each ERN should agree on a detailed action plan to extend the ERN operational disease coverage and develop gradually the specific criteria for each new disease to achieve a comprehensive disease coverage in their respective thematic groupings by 2025.

Target deadline
2022
- 2** Each ERN should match the formal inclusion of a disease with specific networking activities linked to that disease. This would need to be supported by an adequate level of funding to ensure the implementation of these activities.

Target deadline
2022
- 3** The European Commission should launch a call for new Networks to cover the existing disease gaps.

Target deadline
2023
- 4** The Networks should establish cross-ERN working groups for benign tumors and for all the diseases that formally fall under the scope of several ERNs, with a clear annual work plan and measures to assess their work.

Target deadline
2021
- 5** The Networks should enable and consolidate diagonal networking and to integrate health professionals from other disciplines in their collaborative activities on a regular basis. They should establish a cross-ERN working group on integrated care in partnership with European Resources centres for rare diseases to build joint guidance on the provision integrated care.

Target deadline
2023
- 6** All Networks should develop a framework for patient engagement in the ERNs, adequately support the involvement of patient organisations and their representatives in the different ERN activities and fairly compensate patient representatives

Target deadline
2021
- 7** The ERN Board of Member States should develop strategic guidelines on international collaboration and define the role and remit of an “ERN international partner”.

Target deadline
2022



Care

- | | | |
|---|---|--------------------------------|
| 1 | The European Commission, the ERNs and their respective patient advocates, should review the Networks' specific criteria. The EC should conduct a full review of the assessment framework and maintain continuous cycle of independent assessment of full members. | Target deadline
2021 |
| 2 | The European Commission, the ERN BoMS, ERN hospital managers and the ERNs IT WG should agree on a digital roadmap to develop the suite of digital tools and services to support the expansion of cross-border clinical services. | Target deadline
2025 |
| 3 | The European Commission should support the Member States to develop a fair, transparent reimbursement model and establish a funding mechanism for the virtual expert advice consultations and other ERN services under a Networked care model. | Target deadline
2030 |
| 4 | All ERNs should agree and publish a common referral pathway into the Network describing the different referral alternatives and develop information materials. | Target deadline
2022 |
| 5 | All Member States should endorse a hospital as a National Coordination Hub as the central referral management centre to enable access to ERNs specialist advice. | Target deadline
2025 |

Knowledge sharing

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|---|--|--------------------------------|
| 1 | The European Commission should allocate additional direct funding to support the development of new guidelines and other clinical decision support tools (CDSTs), so that by 2025 ERNs will have developed guidance for all rare diseases under their operational scope. | Target deadline
2021 |
| 2 | All the Networks should collect and publish outcome measures that are relevant for patients, for all rare diseases under their operational scope. ERNs must ensure open access to all peer-reviewed scientific publications related to their results and achievements. | Target deadline
2022 |
| 3 | All Networks should hold annual multi-professional and multi-stakeholder clinical summit conferences, to present their outcomes. | Target deadline
2025 |
| 4 | The BoMs, ERNs members and hospital managers should agree on how to facilitate uptake of clinical practice guidelines, CDSTs and clinical pathways at local level. | Target deadline
2022 |
| | Member States, after engaging with the ERNs and professional societies, should validate methodologies for developing clinical practice guidelines, CDSTs and clinical pathways to 'pre-approve' them as ready for implementation at local level. | Target deadline
2025 |

Training and education

- 1 **The Networks should expand their educational and teaching activities**, jointly with Member States, professional societies, rare disease patient community and medical training institutes, **to support the development of local healthcare systems competencies in rare diseases and develop the next generation of experts**, specifically to raise the awareness of rare diseases in medical students of the red-flag symptoms of rare diseases.

Target deadline
2025

- Full Members and Affiliated Partners should establish formal twinning partnerships** to develop the specific knowledge and expertise of the Affiliated Partners.

Target deadline
2021

- 2 **The European Commission should provide more funding to develop innovative training modules and interactive tools to support the ERNs training and education activities**. It should aim to establish an integrated educational strategy for the development of cross-border supra-specialised training.

Target deadline
2025

- 3 **The ERNs Board of Member States should agree on the ERNs strategy to establish education and training agreements with international partners and other clinical networks**.

Target deadline
2025

- 4 **The ERNs should conduct extensive internal training and exchange on clinical governance**.

Target deadline
2025

Research

- 1 **The ERNs Board of member States should agree on the mechanisms to provide sustainable funding to support the operations of the Clinical Research Networks embedded in the ERNs**. Any funding mechanism must ensure the independence of the ERNs to establish their own strategic research agenda.

Target deadline
2021

- 2 **The ERNs should build a common framework for patient engagement into the operations and governance of the individual Clinical Research Networks**.

Target deadline
2023

- 3 **Each ERN should commit to study 80% of the diseases under their operational coverage within a given timeframe**. Over the next two years, each should develop a minimum of 5 new natural history studies.

Target deadline
2022

- 4 **ERNs should develop a comprehensive data strategy and implementation plan** envisaging the necessary activities across 6 action lines: architecture; data collection protocols; data curation services, data management tools, data analytics tools and health data governance.

Target deadline
2022

ⁱ Nguengang Wakap, S., Lambert, D.M., Olry, A. *et al.* Estimating cumulative point prevalence of rare diseases: analysis of the Orphanet database. *Eur J Hum Genet* **28**, 165–173 (2020). <https://doi.org/10.1038/s41431-019-0508-0>

ⁱⁱ EUCERD Recommendations on the Quality of Centres of Expertise, Recommendation 14: Identification of Centres of Expertise

ⁱⁱⁱ European Union Committee of Experts on Rare Diseases, “EUCERD RECOMMENDATIONS QUALITY CRITERIA FOR CENTRES OF EXPERTISE FOR RARE DISEASES IN MEMBER STATES EUCERD Recommendations on Quality Criteria for Centres of Expertise for Rare Diseases in Member States 2,” 2011.

^{iv} EUCERD Recommendations on the Quality of Centres of Expertise, Recommendation 17, 19, 21, 23 and 24: investment in the designated Centres of Expertise to enhance their capacity to deliver these core functions and increase their ability to meet the local population needs and strengthen the foundation of ERNs.

^v EUCERD Recommendations on the Quality of Centres of Expertise, coordination of a specialised multidisciplinary team (EUCERD Q. CoE Rec. 4); building healthcare pathways from primary care (EUCERD Q. CoE Rec. 5); elaborating and disseminating good practice guidelines (EUCERD Q. CoE Rec. 8); providing education and training (EUCERD Q. CoE Rec. 9); developing information adapted to the specific needs of patients and their families, of health and social professionals (EUCERD Q. CoE Rec. 10); contributing to research (EUCERD Q. CoE Rec. 13); liaising with other CoEs at national and European level (EUCERD Q. CoE Rec. 15).

^{vi} C. Hentschker and R. Mennicken, “The Volume–Outcome Relationship Revisited: Practice Indeed Makes Perfect,” *Health Serv. Res.*, vol. 53, no. 1, pp. 15–34, Feb. 2018.

^{vii} J. Morche, T. Mathes, and D. Pieper, “Relationship between surgeon volume and outcomes: A systematic review of systematic reviews,” *Syst. Rev.*, vol. 5, no. 1, p. 204, Nov. 2016.

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RECOMMENDATIONS TO ACHIEVE A MATURE ERN SYSTEM IN 2030

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“Our vision is for a mature ERN system that leaves no person living with a rare disease in uncertainty regarding their diagnosis, care and treatment.”



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