European Commission

Rare Diseases

European Reference Networks

A success story for patients living with a rare disease

European Reference Networks

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European Reference Networks: making a real difference for people with rare diseases

Rare diseases present a unique and urgent challenge in healthcare. All together they affect millions across Europe, and are often complex, poorly understood, and challenging to diagnose and treat. Patients with rare diseases frequently face a long and arduous journey to find the right diagnosis and care, often amplified by the rarity of expertise in their local or national healthcare systems. Moreover, about 95% of the known rare diseases still lack an approved treatment and around 50% lack a confirmed molecular diagnosis.

In the European Union, it is estimated that 6-8% of citizens live with one of the several thousand known rare diseases. This means that around 27 to 36 million people are touched by rare diseases in the EU, which equals the combined population of Belgium and Netherlands or the entire population of Poland.

The European Union has long recognised the importance of addressing rare diseases, taking significant actions in terms of areas of work and financial commitment to promote research, improve access to diagnosis and treatment, improve patient care, and support the development of new treatments, the 'orphan drugs'.

In this context, the establishment of the European Reference Networks (ERNs) marks a groundbreaking milestone in the rare disease agenda, providing tangible added value for patients and their families.

The ERNs were created in 2017 as a collaborative framework that brings together experts and resources from across the European Union: these networks embody the spirit of European solidarity and innovation.

By connecting healthcare providers, researchers, and patient organisations across borders, the ERNs leverage the collective expertise of Europe to ensure that no patient is left behind, no matter where they live, no matter how rare their condition is.

The additional added value of the ERNs lies in their ability to overcome traditional barriers in healthcare. By harnessing advanced digital tools such as an IT platform for cross-border medical discussions on rare diseases using the Clinical Patient Management System 2.0 (CPMS), ERNs enable teams of experts to collaborate on patient cases, offering advice on diagnosis and treatment that might otherwise be inaccessible in a patient's home country.

Furthermore, the ERNs are transforming the research landscape for rare diseases. By pooling data and expertise from across Europe, the networks are accelerating the development of innovative therapies, contributing to clinical trials, and advancing the understanding of rare conditions.

Equally important is the role of ERNs in empowering patients and their families. Through close collaboration with patient advocacy groups, the networks place patients at the centre of their activities, ensuring that their voice and those of their families are heard.

Many actions are currently ongoing in the field of rare diseases, this includes for example:

- revising the EU Pharmaceutical legislation to improve access to safe, more effective, and more affordable medicines, including 'orphan medicines' for patients suffering from rare diseases.
- promoting better cross-border access to health data from rare disease patients via the European Health Data Space.
- implementing Europe's Beating Cancer Plan, which also tackles paediatric cancers, thus complementing the actions implemented by the European Reference Networks focusing on rare cancers.
- reinforcing the work of the 24 ERNs via new direct grants worth EUR 77.4 million. This fund amounts to around EUR 3 250 000 million for each ERN, covering the ERNs' work until September 2027 on consultations, patients' registries, training and clinical practice guidelines, as well as communication activities.
- improving the integration of the ERNs into national health systems and the development of national plans for rare diseases via the Joint Action JARDIN, with funding of EUR 18.75 million.

The work of the ERNs contributes to the completion and the implementation of the European Health Data Space, the pharmaceuticals reform and the Europe's beating Cancer Plan, which are within the priorities indicated in the mission letter to Commissioner Várhelyi.

It is with great pride that I introduce this booklet, a celebration to the dedication, expertise, and vision of those involved in the ERNs. I hope that these stories inspire and reaffirm the importance of continued investment in cross-border cooperation and innovation in the fight against rare diseases. Together, we can ensure that every patient, regardless of their condition, has access to the care and support they need.

Through real-life examples, you will see how these networks have transformed the lives of patients, advanced the frontiers of medical knowledge, and redefined what is possible in healthcare. From groundbreaking diagnoses to pioneering treatments, the impact of the ERNs is far-reaching and profound.

As we look to the future, the ERNs stand as a tribute to what can be achieved through collaboration, innovation, and shared commitment. The ERNs exemplify the power of unity in diversity and the promise of a healthier future for all.

Sandra Gallina

Director-General for Health and
Food Safety, European Commission

What is in this booklet for you?

In this booklet, we provide an overview of the 24 ERNs and celebrate their achievements to the rare disease agenda, to actively communicate on the Commission's actions and decisions and explain the benefits and opportunities stemming from our work.

The overall outlook emerging from reading this booklet is the clear EU-added value that these Networks provide to help patients with rare diseases and their families and the effort that the Commission is making to increase the awareness of the European Reference Networks' activities.

The booklet is non-exhaustive, as the 24 networks provide a truly vast yet diversified array of activities and impacts. More network-specific information is available on the relevant ERN websites, and the Commission encourages readers to continue exploring the ERNs' different activities and outputs.

Table 1: ERNs members (distributed by category)

ERN	Full Members	Associated National Centres	National Coordination Hubs	TOTAL
Endo-ERN	91	13	1	105
ERKNet	64	8	2	74
ERN BOND	44	2	4	50
ERN CRANIO	35	5	2	42
EpiCARE	38	10	2	50
ERN EURACAN	92	7	2	101
ERN eUROGEN	51	1	4	56
ERN EURO-NMD	74	6	2	82
ERN GENTURIS	44	5	2	51
ERN GUARD-Heart	43	7	2	52
ERN PaedCan	79	9	2	90
ERN RARE-LIVER	52	7	3	62
ERN ReCONNET	54	6	3	63
ERN RITA	61	7	2	70
ERN TRANSPLANT-CHILD	33	4	3	40
ERN-EuroBloodNet	90	4	3	97
ERN-EYE	51	5	3	59
ERN-ITHACA	66	2	3	71
ERN-LUNG	78	7	2	87
ERN-RND	63	2	2	67
ERN-Skin	52	2	2	56
ERNICA	39	9	4	52
MetabERN	85	4	2	91
VASCERN	39	4	2	45
TOTAL	1 418	136	59	1 613

The European Reference Networks

The European Reference Networks (ERNs) are cross-border networks that bring together European hospital centres of expertise and reference to tackle rare, low prevalence and complex diseases and conditions requiring highly specialised healthcare.

ERNs enable specialists in Europe to discuss cases of patients affected by rare, low-prevalence and complex diseases, providing advice on the most appropriate diagnosis and the best treatment available.

Individual patients cannot directly access ERNs. However, with the patient's consent, healthcare providers can exchange information and consult the appropriate ERN under national health systems.

There are currently 24 ERNs created under Directive 2011/24/EU on patients' rights in cross-border healthcare. They include 1 613 specialised centres located in 382 hospitals across 27 Member States and Norway.

The ERNs carry out several activities, and includes, for example:

- **Development of Clinical Practice Guidelines (CPGs):** the ERNs develop, update, and appraise Clinical Practice Guidelines, which are recommendations for clinicians about the care of patients with specific conditions. They are based on the best available research evidence and practice experience.

- **Development of care pathways and patients' journey:** A care pathway is a complex intervention involving mutual decision-making and organisation of care processes for a well-defined group of patients over a well-defined period. A patient journey refers to the needs and experiences of patients and caregivers from their clinical presentation through key stages of their healthcare journey. It encompasses both medical and psychological aspects. Each rare condition has a unique progression and course, from its onset through its lifespan. Therefore, it is important to develop a tailored set of care pathways and patient journeys for each rare condition.

- **Training and Education activities:** the ERNs provide training and educational courses for their members through webinars, conferences, clinical exchanges and endorsement/accreditation of educational activities. Several materials are available on the ERNs' websites for further dissemination and use.

- **Virtual Expert Discussions:** European specialists discuss cases of patients with rare, low-prevalence, and complex diseases, providing advice on diagnosis and treatment. These discussions are facilitated by the Clinical Patient Management System (CPMS), an IT platform in place since 2017, which has been recently upgraded to be a more user-friendly, secure, GDPR-compliant, and open-source version, the CPMS (2.0).

- **Registries:** the ERNs have set up transnational registries to collect pseudo-anonymised data on patients with rare diseases. They are an important component of digitalised European healthcare establishing a rare disease registry ecosystem based on interoperability and FAIR[1] principles. They harmonise data on rare disease patients across the EU, and make data collected available to researchers, public authorities, industry and other stakeholders on different conditions to improve the medical care of patients.

- **Research activities:** ERNs are involved in a few clinical trials and prioritise unmet research needs in the field of rare diseases to put the patient voice at the core of research needs. To learn more, visit Research and innovation on rare diseases.

1 Findable, Accessible, Interoperable, Reusable

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Patient Stories

The ERNs have multiple examples of real-life patient stories where they have been able to directly and positively influence the outcome for patients and families. Below are a few excerpts to highlight this work.

Anna, a young woman in Italy, was constantly tired and short of breath. She couldn't run, She couldn't participate in sports. She had no energy. She was diagnosed with pulmonary arteriovenous fistula, a rare condition often associated with hereditary haemorrhagic telangiectasia (HHT) and typically present from birth, in which blood bypasses the lungs and is not replenished with oxygen. In many cases, there are no symptoms, which can be dangerous because it can lead to blood clots.

An accurate diagnosis and specialised care can be hard to get. Luckily, experts at at the VASCERN Hereditary Haemorrhagic Telangiectasia Centre in Crema (Italy), ensured she got the correct care. Following their advice, Anna had embolization surgery and recovered so well that she even won a running competition soon after!

In another case, **a little girl developed a very early onset of puberty due to an ultra-rare disease of a pituitary stalk tumour producing Human Chorionic Gonadotropin (HCG).** She needed immediate treatment, but her insurance denied it because no biopsy had been taken beforehand.

Specialists working with Endo-ERN said chemotherapy should be started immediately, and that conducting a biopsy might endanger the child and delay the treatment. The health insurance accepted the expert opinion.

Within a week of the tumour's discovery, the patient began treatment. Not only was this a happy ending for one young girl, but it may also help others in the future - a precedent was set not to delay oncology treatment if the tumour is otherwise well characterised and if the biopsy itself could endanger the patient.

This is the story of Justine from France, who says she was born twice, on her birthday and the day of her transplant. Justine was just 3 and a half years old when she had a double transplant, and it was the first of its kind in Europe. Today, 28 years later, she knows how lucky she was and encourages parents to stay positive and children not to be afraid.

Another one is from **Fernando from Spain, who had renal failure and a liver transplant at age 14.** He's still 14 and says that life is great without dialysis and it's a miracle to be able to eat 'almost' normally again.

Another one comes from a mother in Italy who took her three-year-old to a doctor while on vacation, thinking her child had eaten some bad fish. The doctor recognised something else was happening, and in less than 24 hours, the little girl was diagnosed with a rare liver cancer. Today, she is an energetic 11-year-old. 'Kids teach us not to give up and always fight,' says her mother.

Patient Partnerships and ERNs

The patients themselves are always at the heart and centre of the ERNs. Their integration and participation ensure that patient voices are heard and reflected in the ERNs' work.

ERN RARE-LIVER, which works on rare liver diseases, provides an example of its RARE-LIVER Youth Panel and highlights how it involves young patients as mentors, role models, and collaborators in its work.

As anyone over the age of 30 knows, young people tend to speak their own language and strongly identify with their peers. So, who better to serve as mentors and role models for young patients with rare liver diseases than young people who are going through some of the same experiences?

The RARE-LIVER Youth Panel comprises 10-15 people between the ages of 18 and 30 living with rare liver diseases. This group meets at least six times a year online and once a year in person. The members also closely follow the activities of ERN RARE-LIVER, participate in workshops and online meetings, and help develop guidelines.

> "We focus on improving medical care for young patients and strive to make their journeys easier by providing support and connection"

By sharing their experiences with health professionals, panel members help improve healthcare and provide valuable input from the patient's viewpoint and from a young person's perspective. Mentoring other young people with rare liver diseases provides an invaluable service—it helps these young patients feel less afraid, more optimistic, and understood.

Panel members also benefit from the opportunity to talk amongst themselves and with other young people in the rare disease community and make positive changes in healthcare and in the lives of young people going through their own rare disease journeys.

ERN CRANIO, which works on rare and/or complex craniofacial anomalies and ear, nose and throat disorders, presents a project, 'With my family to the theatre', to help look at the understanding between care professionals and patients/caretakers. The question at the heart of this project was, "How can we, as doctors and healthcare providers, be an even better guide for our patients by seeing every aspect of their lives, and not just the physical part? And by showing more of ourselves as human beings"? The project resulted in a film that explores the role of parents and families for patients with craniofacial conditions and the need for good doctor-patient relationships for more effective treatment and measurable health benefits. The film explores this relationship and brings to the fore the need to raise attention to the doctor-patient-parent relationship.

The ERN EYE launched a serious game on best practices to announce a diagnosis.

Inspired by real life situations, this project allows users to confront difficult situations when delivering a complex diagnosis.

The development of this project brought together a committee of medical experts as well as patient associations and psychologists.

Access the Serious game here:
ERN-EYE Serious Game - ERN-EYE

Patient Journeys

One of the most frightening things a patient faces is the unknown. What symptoms will they have? What changes can they expect? How will they feel physically and emotionally as they go through treatment for their illness? Patient Journeys provide this kind of information from the patient's perspective, providing not only vital information but also reassurance.

ERN-RND, working on rare neurological diseases, provides an example of specific Patient Journeys in the form of easy-to-understand factsheets for five disease groups and is developing three more to cover other groups. These Patient Journeys are co-produced by patients and clinicians and represent a step forward toward systematic patient engagement in the design of care pathways

The goal is to hand the appropriate Patient Journey to each newly diagnosed patient to help answer their questions and help them feel less fearful, supported and more empowered.

Watch the videos:

- **Patient Advocate Lori Renna Linton telling her own HSP story: https://www.youtube.com/watch?v=NHpGRGHfQlc**
- **Patient Advocate Juliane Krabath, who has a child with HSP: https://www.youtube.com/watch?v=nYSy99xYoZA**

Disclaimer: If you have any concerns or questions about the use of your personal data in YouTube, you should read its privacy policies carefully.

Two patient advocates share how the Patient Journeys on Hereditary Spastic Paraplegias (HSPs) have been helpful

Patient advocate Lori Renna Linton has had HSP for 16 years. she says her life would have been so much easier if she'd had a Patient Journey available earlier on. Thinking back to when she had his appendix removed in 2017, she says the Patient Journey could have helped her convince her healthcare providers that HSP was a real disease and show them that it's Googleable. It would have told the nurse on duty that the black sweatpants with the white stripe down the side did not mean that she was a kickboxer. It would have said that she gets cramps in her feet and legs. "But the biggest win of all is that the Patient Journey would have allowed me to get the proper care needed and that I would not have to repeat my story over and over and over," she said.

Patient Advocate Juliane Krabath, the mother of a 14-year-old girl who was diagnosed with HSP when she was 8, said that at the beginning, the family didn't know much about HSP. But a Patient Journey told them they could expect a constant worsening of symptoms, especially impacting their daughter's ability to walk. The girl attended a normal Austrian school, which was not entirely accessible. "At first, this was no problem, but now, four years later, she needs walking sticks, and the changes from classroom to classroom get more and more exhausting to her." Having the Patient Journey helped them make the decision to change to a more accessible school that was better connected to public transport. "Having a patient journey that shows the progress of the disease and the challenges that come up could help other families to prepare better for the future too", she said.

Patient Journeys are available in the main European languages and available as flyers at:
https://www.ern-rnd.eu/disease-knowledge-hub/ern-rnd-patient-journeys/

Patient's pathways and promotion of early diagnosis

ERN ReCONNET promotes early diagnosis and improved organisation of care by developing patients' pathways and a diagnostics tool, Red Flags, adopting a multi-stakeholder approach.

ERN ReCONNET designed a methodology (**RarERN Path™**) that can be applied in all rare and low prevalence diseases. It aims to create a reference organisational model for patients' care pathways which, if applied in different contexts, helps to ensure improved, cost-effective and patient-centred equal care for rare and complex diseases.

RarERN Path: a methodology towards the optimisation of patients' care pathways in rare and complex diseases developed within the European Reference Networks. https://doi.org/10.1186/s13023-020-01631-1

The methodology provides for the involvement of different stakeholders, such as medical experts, health economists, patient representatives, caregivers, clinicians from different specialties, and other healthcare providers (nurses, psychologists, etc.). It has already been applied for different diseases within the remit of ERN ReCONNET (e.g. systemic sclerosis).

At the same time, ERN ReCONNET is developing Red Flags for the early diagnosis of rare and low prevalence connective tissue and musculoskeletal diseases. The first Red Flags tool was developed for one of the rarest conditions covered by ERN ReCONNET, the IgG4-related diseases. The Red Flags are developed using a multi-stakeholder approach involving medical experts, patient representatives, and general practitioners. The dissemination of these Red Flags in hospitals and academic environments aims to increase disease awareness among specialists who are not experts in the specific disease, as well as among medical students.

More information:

Improving organisation to improve care: ERN ReCONNET organisational reference model for systemic sclerosis patients' care pathway
https://doi.org/10.1177/23971983241269109

An Opportunity to Harmonise the Approach to Patients' Care Pathways for Rare and Complex Diseases: RarERN Path™
https://doi.org/10.3389/frhs.2022.935014

Identification of red flags for IgG4-related disease: an international European Reference Network for Rare Connective Tissue Diseases framework
https://doi.org/10.1016/s2665-9913(24)00192-9

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Emergency cards

In this story, ERN-Skin has provided a tailored solution for patients with rare skin diseases who may need urgent care when away from home. So, what does happy when a rare skin disease patient finds themselves in an emergency situation and ends up in urgent care? What should they do?

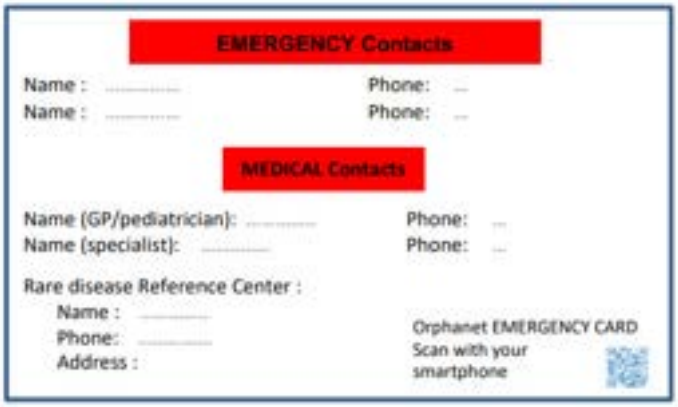
The answer? Carry a special emergency card, since most medical staff will be unfamiliar with rare skin diseases.

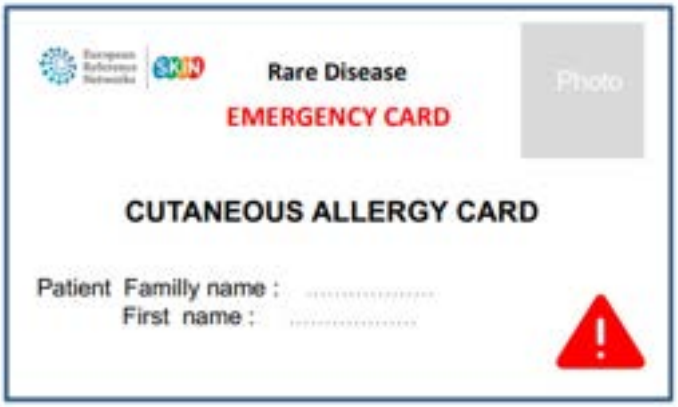
Produced by the ERN-Skin experts, along with SKIN European Patient Advocacy Groups advocates, emergency cards have all the essential information on how to manage a patient with a rare skin disease. In an emergency situation, it is vital to provide this adapted care to avoid any complications.

These emergency cards are the size of a business card and are written in the national language, so that patients can carry them at all times.

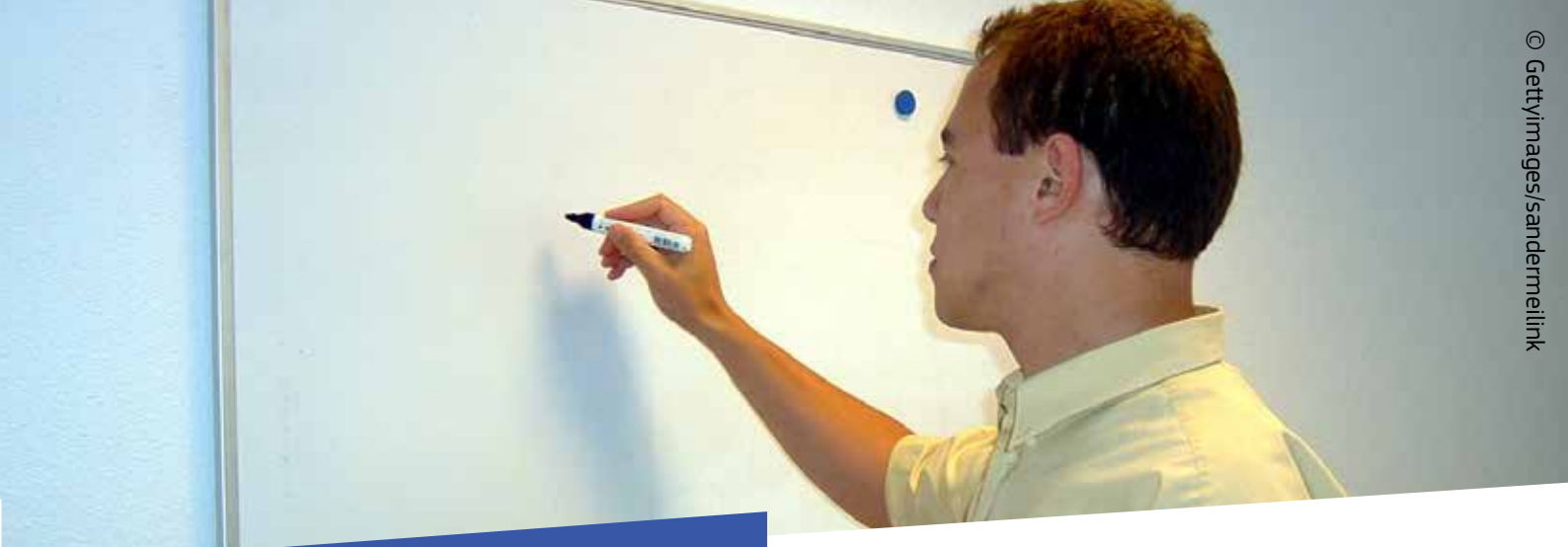
ERN-Skin has developed emergency cards for the following diseases:

- Ectodermal dysplasia
- Epidermolysis bullosa
- Ichthyosis
- Severe cutaneous drug reactions





More information is available at: https://ern-skin.eu/emergency-cards/



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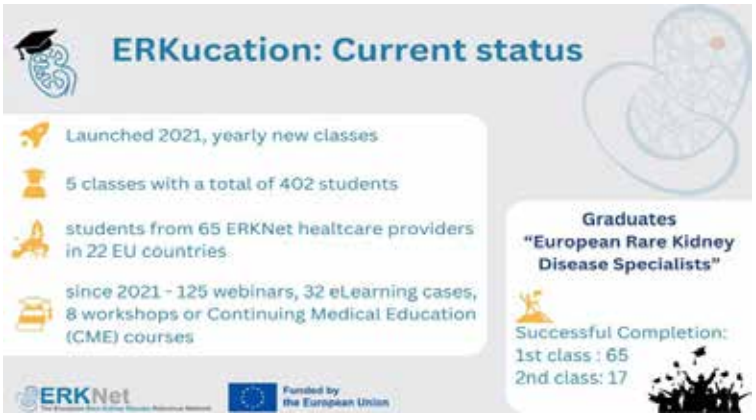
First ERN postgraduate curriculum in the field of rare kidney diseases (ERKucation)

Education and training are essential activities for all ERNs, strengthening and improving the future healthcare professionals specialising in rare diseases and complex conditions across Europe. Academic curricula on rare diseases are not so common, making the development of such programmes an important task of the ERNs.

ERN ERKNet highlights such an example in the field of rare kidney diseases.



The first ERKNet class graduated in December 2023 with 65 European Rare Kidney Disease Specialists.

ERN engagement during the COVID-19 emergency

The ERNs also quickly reacted with their support when the COVID-19 pandemic emerged. Below are a few examples of ERN emergency engagement to help the EU response to COVID-19.

To help rare bone disease patients get the support and care they needed during the pandemic, the ERN BOND and several Italian healthcare professionals set up a direct and dedicated 24/7 telephone line, "COVID-19 Helpline for Rare Bone Diseases". The aim was to share experiences and knowledge about rare bone diseases with both the patients themselves and healthcare professionals working in the intensive care units and/or COVID-19-wards treating patients affected by rare bone diseases. For rare bone disease patients, it was crucial to know that they could still be in close contact with their care physicians and keep them informed about any changes in their health.

ERN ReCONNET, working on Rare and Complex Connective Tissue and Musculoskeletal Diseases (rCTDs), also sprang into action and produced points to consider for treating patients living with autoimmune rheumatic diseases with antiviral therapies and anti-SARS-CoV-2 antibody products.

This came alongside the design of a multicentre prospective cohort study dedicated to COVID Vaccination in rare and complex connective tissue diseases (VACCINATE) and a series of workshops to define the challenges on the management and vaccination of rCDTs patients in the COVID-19 era as well as informative posters on vaccination al EU languages.

This successful experience highlighted the fundamental role of remote high quality of care for rare bone diseases during the COVID-19 outbreak that could become a **gold-standard practice for remote care**, particularly relevant for rare bone diseases patients.

More information:

The line between COVID-19 pandemic and rare bone diseases
https://doi.org/10.1007/s11845-020-02400-6

Providing high-quality care remotely to patients with rare bone diseases during COVID-19 pandemic
https://doi.org/10.1186/s13023-020-01513-6

ERN ReCONNET points to consider for treating patients living with autoimmune rheumatic diseases with antiviral therapies and anti-SARS-CoV-2 antibody products
https://doi.org/10.55563/clinexprheumatol/jpargp

The impact of COVID-19 on rare and complex connective tissue diseases: the experience of ERN ReCONNET
https://doi.org/10.1038/s41584-020-00565-z

Long-term outcomes of COVID-19 vaccination in patients with rare and complex connective tissue diseases: The ERN-ReCONNET VACCINATE study
https://doi.org/10.1016/j.jtauto.2023.100221

Dedicated Rare Disease Expertise

ERNs work with patients and care team members. ERN LUNG, which works on rare respiratory diseases, described a unique system for answering questions in their dedicated field.

The creation of the ERN-LUNG Expert Advisory BOard (ERN-LUNG EXABO) was set up primarily for patients and care team members for any ERN-LUNG related questions. As of 31st of August 2023, 35 new patients have used the system to enquire and get their questions answered by rare disease experts.

The system is expanding its availability in European languages and has the strong support of patient organizations who are helping to spread the word about this innovative system. Moreover, the ERN has considered a step-by-step process; if the answer cannot be provided using this, it goes upwards for a medical discussion in the Clinical Patient Management System.

Multidisciplinary expert approaches

Multidisciplinary expert approaches, such as Multidisciplinary Tumour Boards, are fundamental to achieving holistic and comprehensive reviews of patient diagnosis and treatment. ERN EURACAN, which works on rare adult solid cancers, provides an example of how this approach is integrated into its work and the added value it provides for patients.

ERN EURACAN highlights the example of the rare gynaecological cancer group, under which 260 patients' cases were reviewed between November 2017 and October 2023.

Impact of Multidisciplinary Tumour Boards (MDTBs) on patient care[2]:

Since March 2022, as part of EURACAN strategy to harmonise activities between all rare solid cancer groups, the ERN seized the opportunity to support virtual MDTBs with a specific funding included in the EU grants and dedicated to the use of the Clinical Patient Management System (CPMS).

- These MDTBs gather leading EURACAN experts from different countries to review complex or very rare patient cases in all cancer groups and to consider all perspectives and give timely and accurate diagnoses.
- These online discussions are also meant to increase access to novel treatments and clinical trials.

Each case, in each cancer group, is then entered on the CPMS to develop the use of the platform across the Network.

2 Alice Bergamini, ESMO Gynaecological Cancers Congress 2024 #ESMOGynae24, Abstract 82MO

- The number of reviewed patients doubled over 6 years.
- Further diagnostic testing in 1/3 of patients.
- Alternative treatment opportunities to those initially planned for 50% of patients.
- 94% adherence to these treatment recommendations.
- Surveillance, instead of adjuvant chemotherapy, was recommended in 17% of patients.
- 37 patients gained access to off-label therapies, 4 were enrolled in clinical trials abroad.

The MDTBs allowed patients to access off-label therapies not yet approved for rare gynaecological cancers, which would otherwise not have been accessible in some countries.

Inter-ERN cooperation

ERNs often face the same challenges, which they can tackle more efficiently together! Here, you can read how two ERNs – ERN eUROGEN and MetabERN – are working together to help speed up the development of paediatric and orphan devices for rare diseases.

In the field of rare diseases, there aren't enough medical devices specifically developed for children with rare diseases. These devices, called paediatric and orphan devices, can provide essential functions for patients with rare diseases, their carers, and the healthcare professionals using them, yet many patients and carers do not have paediatric devices that are tailor-made for their conditions.

That's why the DeCODe platform initiative is working to encourage the development of paediatric and orphan devices, and it will do this in two steps:

- DeCODe will first map paediatric and orphan medical devices, stakeholders and initiatives and will then develop a critical pathway analysis to determine the optimal way to develop novel paediatric and orphan medical technologies.
- Then, DeCODe will select five developers to support. For each of the successful developers, DeCODe will keep track of the funded paediatric orphan devices, the number of prototypes that each developer has developed, the business plans that are drafted, the number of clinical data collections that have been launched, and certificates that have been obtained throughout the support process.

The expected result? This new initiative will approve and implement 3-5 new medical devices for children in rare disease care.

The DeCODe platform will be available for any European developer (a patient-led group, academic, or small to medium enterprise).

And you can read here about how EURO-NMD, ERN-RND, and EpiCARE work together on gene therapy practices and on mitochondrial disease care and research

The inter-ERN Working Group on Gene therapies involves 3 ERNs (ERN EURO-NMD, ERN-RND, EpiCARE), with the aim of working together on actions addressing aspects of gene therapy that are transversal to the 3 ERNs, e.g. current practices, regulatory and healthcare cost-related challenges, clinical trials, etc. In this context, a series of webinars on gene therapies took place, sharing lessons learned and practical implications.

In addition, a large survey was conducted to assess the current landscape of approved and investigational gene therapies in Europe. ERN-EYE and MetabERN were invited to participate. The analysis is ongoing and its outcome should be a position and recommendation paper to improve and harmonize current practices around gene therapies across Europe.

The Mito inter-ERN Working Group involves 5 ERNs (ERN EURO-NMD, ERN-RND, EpiCARE, ERN-EYE, MetabERN), with the aim to address cross-diseases challenges around care, education and research on Primary Mitochondrial Diseases (PMDs). In this context, 24 European experts have worked together to develop guidelines and consensus recommendations on safe medication use and seizure management in mitochondrial epilepsy. The outcome of this collaborative work was published in the European Journal of Neurology https://doi.org/10.1111/ene.16275/

DeCODe

More information about DeCODe can be found here: https://eurogen-ern.eu/decode-orphan-and-paediatric-medical-device-project-launched/

© Gettyimages/MJ_Prototype

Ensuring drug sustainability

Another challenge often faced in the field of rare diseases is the availability and access to specialised medicines and treatments for rare diseases. MetabERN, which works on hereditary metabolic disorders, describes how they overcame a shortage of a lifesaving drug.

MetabERN explains that Cobalamin C defect (CBLC) is a rare congenital disease affecting the metabolism of vitamin B12 (cobalamin), which is lethal if not treated. There is only one known treatment - the daily administration of high-dosage hydroxocobalamin (OHB12). Without this drug, patients with CBLC suffer serious medical complications and even die.

So, what could patients do when the marketing of this life-saving therapy was discontinued in 2022, and shortly afterwards, shortages of OHB12 were reported? Their lives were literally at stake.

In response to this shortage, Italian associations, organisations, and medical professionals took action. The Associazione Italiana Acidemia Metilmalonica con Omocistinuria (the Italian Methylmalonic Acidemia with Homocystinuria association) and the MetabERN Italy team, led by the Bambino Gesù Children's Hospital in Rome, joined forces with other patient organisations, clinicians, and the Italian Medicines Agency.

After analysing different options, MetabERN Italy concluded that no other option could be found: patients needed OHB12, which could only be imported in limited amounts from Spain.

As a result, the Military Pharmaceutical Chemical Institute of Florence, Italy, tasked by law to find solutions to national emergencies, stepped in to make new stocks of OHB12 available for all families needing treatment. The supply was restored, saving patients' lives.

Working with Orphanet

High-quality information and accurate rare disease classification are essential for the identification, diagnosis, care and treatment of rare diseases. ERN ITHACA, which works on rare malformation syndromes, intellectual and other neurodevelopmental disorders, explains how they cooperate with Orphanet in this area.

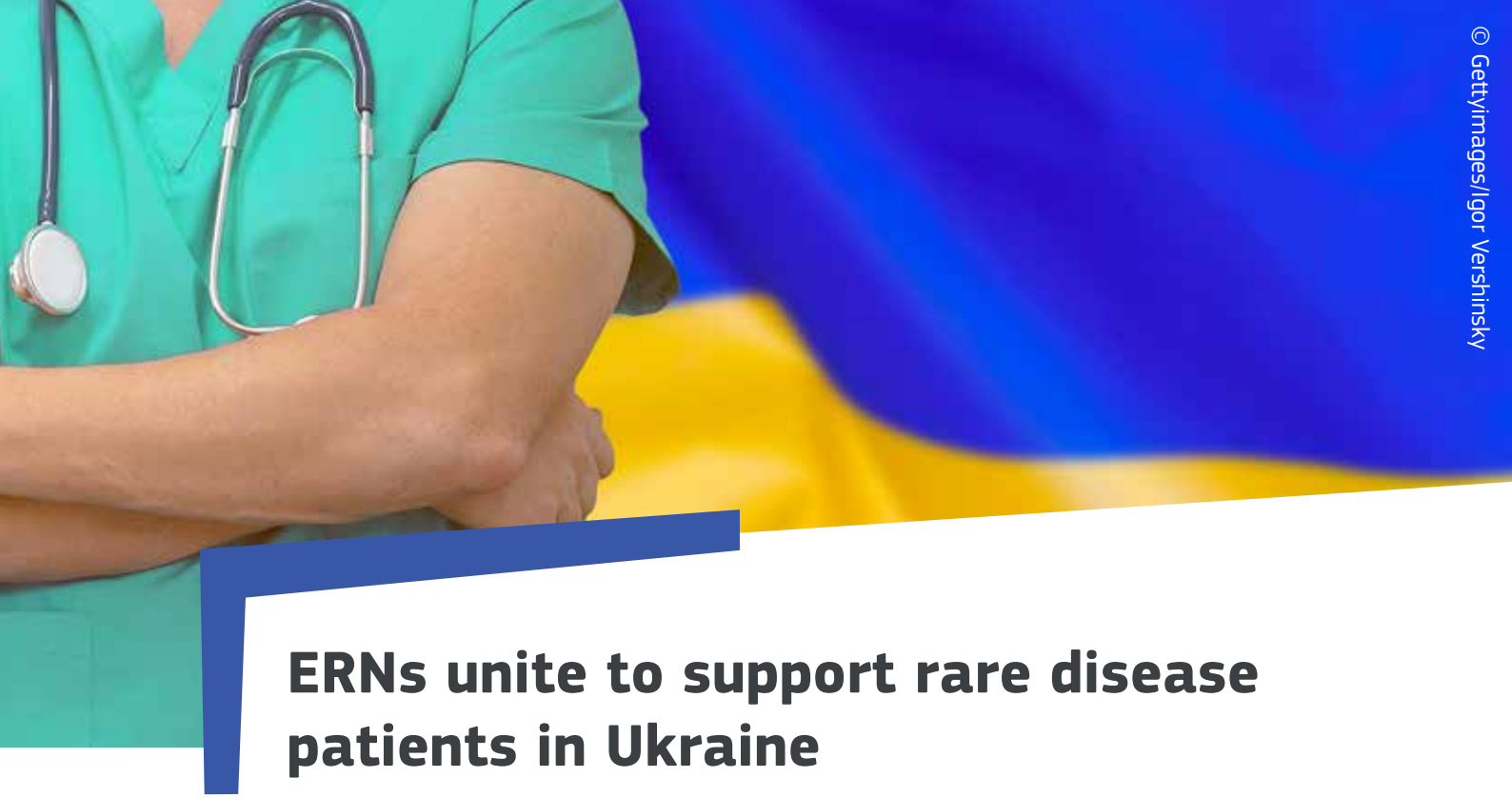
Orphanet, the portal for rare diseases and orphan drugs, is a digital platform that:

- gathers, improves, and provides high-quality information on rare diseases to improve the diagnosis, care and treatment of patients with rare diseases.

- maintains and develops the Orphanet rare disease nomenclature (ORPHAcode), an internationally valid classification system for rare diseases that serves to identify rare disease patients, which the European Commission also recommends.

Every participating healthcare provider in the ERN ITHACA network is invited to collaborate with Orphanet to create or update entries in the European catalogue for (neuro)developmental disorders. Over 80 contributions have been uploaded online since 2020. This information can be fed into the SysNDD database, for example, a database of genes involved in intellectual disability and autism that ERN ITHACA supports and helps curate. ERN ITHACA also funds the link between Orphanet and the database for the list of non-syndromic ID genes.

ERN ITHACA has also contributed to several updates in the ontology used by ORPHANET, based on the Human Phenotype Ontology (HPO) database. The main contributions are in the field of foetal pathology as well as recent updates in the classification of spinal dysraphisms, the latter which was done in collaboration with ERN eUROGEN.



ERNs unite to support rare disease patients in Ukraine

In March 2022, the European Reference Networks sprang into action to support the more than two million Ukrainian patients estimated to have a rare or complex disease. Already in the first three months of Russia's war of aggression against Ukraine, almost 300 cases were referred to ERN Healthcare Providers, most of them patients with rare paediatric cancers, blood disorders, or developmental diseases. In addition, the Commission established a framework that allows Ukrainian healthcare providers to seek advice on Ukrainian rare or complex disease patients from members of the European Reference Networks.

ERNs are built on the principle that medical expertise should come to the rare disease patient, and not the other way around, and nowhere is that more crucial than in a war-torn country. Complicating the already complicated challenge of providing accurate diagnosis and care for a rare disease were needs that specifically arose due to the conflict. Patient files were destroyed or lost, there was a sudden need for translation services, and there was a shortage of facilities and healthcare providers.

Other specific medical needs included:

- Support on how Ukrainian patients can access highly specialised surgeries or medical devices.
- Support to healthcare professionals in Ukraine and patient associations.
- Support in triage and referral of refugees to ad hoc EU medical teams.
- Support connections between patients and associations, such as EURORDIS.

Several ERNs implemented activities to support Ukrainian rare disease patients, such as:

- As of September 10, 2024, the European Paediatric Oncology Community, including ERN PaedCan, on paediatric cancer (haemato-oncology), assisted 1 643 Ukrainian paediatric cancer patients through the SAFER (Supporting Action for Emergency Response) Ukraine programme.

- After the attack on the Okhmatdyt National Children´s Hospital in Kyiv, Ukraine, on July 8th, 2024, paediatric cancer patients needed to be evacuated again. ERN PaedCan members closely collaborated with SAFER Ukraine and/or directly with European National Health Ministries to safely evacuate 13 patients.

- EpiCARE, the European Reference Network on rare and complex epilepsies, is officially represented at the "Emergency and Crisis Response Task Force" for Ukraine, created by the International League Against Epilepsy (ILAE). The group agreed on a set of seven key actions. Monthly calls and direct contact with epilepsy experts and patient advocates in Ukraine are ensured by the ILAE Task Force and the ERN EpiCARE to oversee implementation and update these actions. A webinar on Managing Epilepsies in Crisis Situations – The Ukraine Experience, with invited speakers from Ukraine, was recently released by ERN EpiCARE.

- Just after the military assault at the end of February 2022, under the initiative of ERN-EYE, the ERNs created a dedicated website, https://www.erncare4ua.com/, to provide information on each of the 24 ERNs and general information and news in English and Ukrainian. The website includes information on how to find a centre in the EU for a rare or ultra-rare disease, how to find medical support for diagnosis, how to find medical support for treatment issues (drugs, surgery, etc.…), how to find medical support for medication changes or shortages, and how to find other clinically tailored help via a general contact point.

New Guidelines and Recommendations

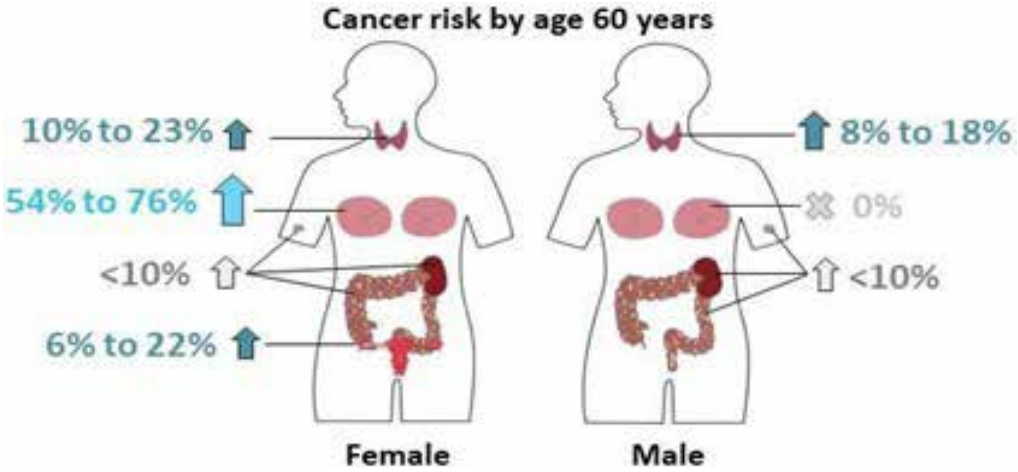
ERN GENTURIS sets new guidelines for cancer screenings for patients with PTEN hamartoma tumour syndrome, offering them earlier and more frequent screenings for earlier detection and better outcomes.

The possibility of getting cancer scares most people, but for those with a high hereditary cancer risk, living with that fear can rob them of some of the joy in life by causing stress and anxiety. Patients with rare genetic tumour risk syndromes, are at a very high hereditary risk of developing common cancers. For instance, people with PTEN hamartoma tumour syndrome (PHTS) are at an increased hereditary risk of breast, thyroid, endometrial, renal and colorectal cancers. If they do get cancer, they need different treatments and follow-up compared to other cancer patients.

For PHTS patients, one way to reduce stress about cancer is to get regular surveillance. If cancer is caught early, it can be more easily treated, and the outcomes are much more likely to be favourable.

The new ERN GENTURIS guidelines recommend earlier and more frequent screenings than those for the general population. For example, after the age of 18, patients should have an annual ultrasound test to check for thyroid, and after age 30, they should have an annual MRI to check for breast cancer.

No one enjoys going in for tests that reveal a problem. But if there is a problem, it's always best to tackle it when it's small!



The ERN EuroBloodNet holistic approach to address Sickle cell disease (SCD).

ERN-EuroBloodNet, dedicated to rare hematological diseases, has developed a holistic approach to address sickle cell disease (SCD) a hereditary life-threatening disease. Coordinated actions across EU Member States involving healthcare experts, researchers and patients advocates, have been implemented. It involves done and ongoing actions and is a model to address other rare or ultra rare haematological diseases.

ERN-EuroBloodNet's contribution to the SCD Lancet Commission "Defining Global Strategies to Improve Sickle Cell Outcomes" is a testament to this.

More information:

Sickle cell disease landscape and challenges in the EU: the ERN-EuroBloodNet perspective
https://doi.org/10.1016/s2352-3026(23)00182-5

The added value of using the Clinical Patient Management System (CPMS)

Using the CPMS for carrying out virtual consultations of patients in the EU and Norway can make a real difference for the patient, as it provides a means for expert diagnosis and treatment. This is illustrated by the following stories involving patients of some ERNs.

ERN-Skin

During CPMS discussions between ERN-Skin experts about a Dystrophic Epidermolysis Bullosa patient with unexpected severe cardiac distress, it was discovered that isolated cases had been observed within several ERN-Skin clinical units.

ERN-Skin experts decided to group these cases to better understand them. This led to a **recommendation for systematic cardiological monitoring** and a **research project** to improve patient management (frequency of cardiac complications during Dystrophic Epidermolysis Bullosa, predisposing factors).

The recommendation was then discussed at the international level during the 2nd World Congress on Rare Skin Diseases, co-organised by ERN-Skin in Paris, which gathered more than 500 participants (medical doctors, researchers, patient representatives, students, the industry) from 58 countries.

ERN RITA

A medical doctor from an ERN RITA healthcare provider was treating a patient with a challenging refractory disease (dermatomyositis) and turned to the CPMS for advice.

The rapid CPMS interaction with ERN RITA specialists across Europe led to the identification of a more effective therapy for the patient.

ERN GUARD-Heart

ERN Guard Heart explains the use CPMS to improve participation of cardiac rare disease patients in sports: Long QT syndrome (LQTS) is a rare heart condition associated with life-threatening arrhythmias and an increased risk of sudden cardiac death at a young age. Physical activity and emotional stress are known triggers of arrhythmias and sudden cardiac death, but not all LQTS patients are affected, and it remains unclear how to identify those at higher risk.

This uncertainty causes anxiety among patients and healthcare providers, leading to inconsistent advice regarding sports participation.

To address these discrepancies and provide clearer and more consistent guidance, ERN GUARD-Heart has formed a multidisciplinary advisory panel of European experts in LQTS.

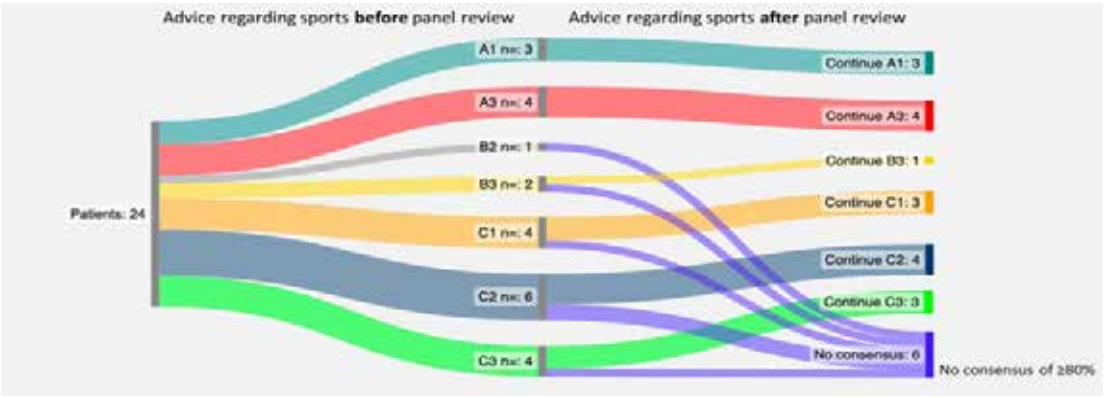
Healthcare professionals across the EU can consult this panel through the CPMS by submitting cases of LQTS patients who participate or wish to participate in sports. The panel reviews each case and provides recommendations addressing the following key questions:

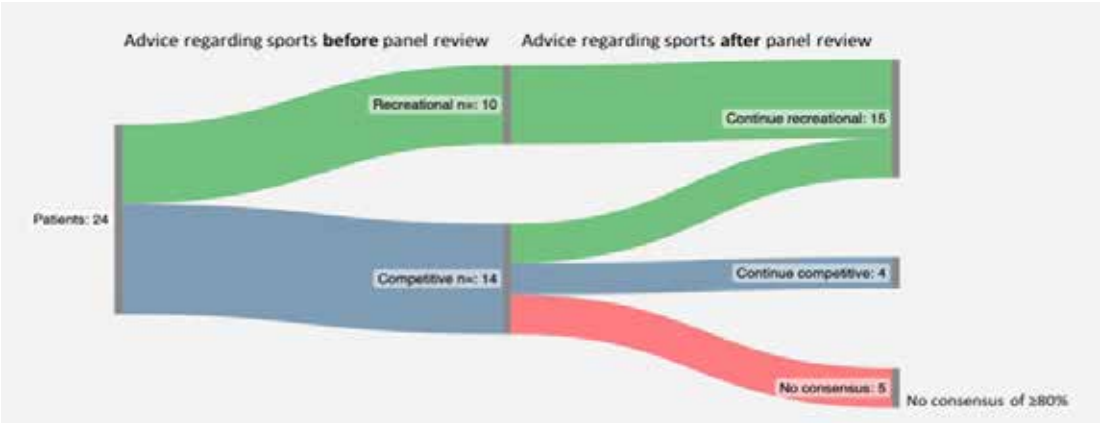
- Should the patient limit or restrict his/her participation in sports?
- Which types of sports are permissible based on the sports categories displayed in Table 1?

Data from these consultations are used for research and educational purposes to improve care and ensure safer sports participation for LQTS patients.

The recommendations for the first 24 patients discussed in the CPMS are summarised below.

Table 1: Classification of organized sports and exercise.



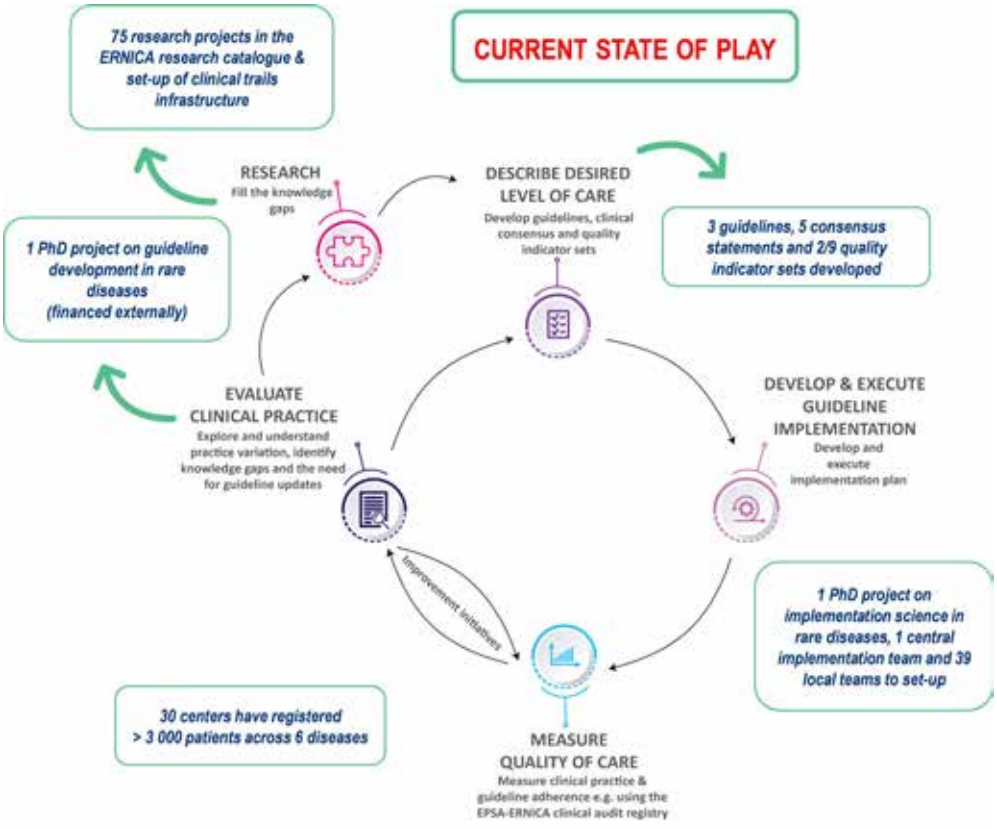


Quality cycles

The ERNs connect hospital centres of expertise and reference and work hard to ensure the accuracy of diagnosis and quality of treatment.

ERN ERNICA, which works on rare inherited and congenital anomalies, has developed its own quality cycle to share knowledge and ultimately care for and cure patients. Quality cycles outline the four stages of continuous improvement, often categorised as plan, do, check, and act.

This involves developing and implementing clinical guidelines, evaluating the patient registry, and filling knowledge gaps in research (an ongoing process).

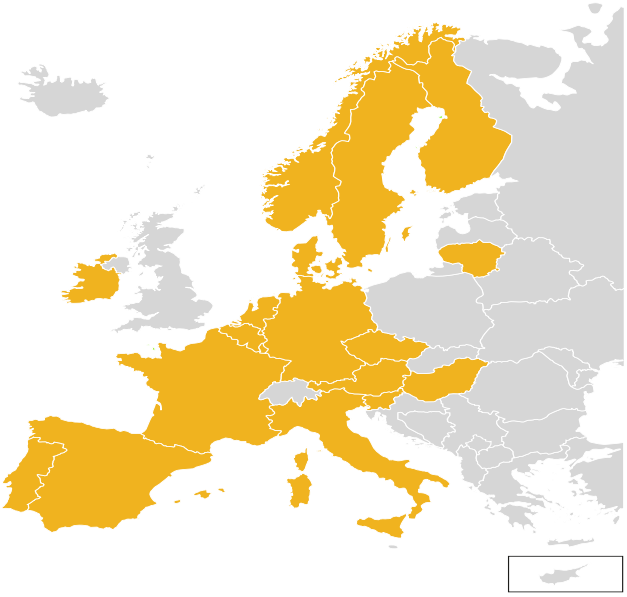


ERN Facts and Figures

ERN BOND

European Reference Network on rare bone disorders

https://ernbond.eu/



Members

50 healthcare providers located in 19 countries in the EU and Norway are members of the network, distributed as follows:

- 44 Full Members in 13 Member States, plus Norway
- 2 Associated National Centres in 1 Member State
- 4 National Coordination Hubs in 4 Member States

In addition, 9 European Patient Advocacy Groups (ePAGs) and other national patient organisations involved in ERN activities are collaborating

Disease areas

- Skeletal dysplasias and metabolic bone-related conditions and all rare diseases affecting cartilage, bone, and dentine.
- 771 different rare bone disorders associated with 552 genes, classified into 41 groups (according to the 2023 Nosology of genetic skeletal disorders)

Guidelines, care pathways, and patient journeys

- 1 guideline currently under drafting by ERN BOND
- 26 guidelines authored and endorsed by ERN BOND
- 2 patient journeys in progress

Training and education

- 22 webinars organised by ERN BOND since 2021, and 6 in collaboration with the European Rare Disease Research Coordination and Support Action (ERICA)
- 6 short exchange programmes
- 13 thematic workshops

Clinical Patient Management System (CPMS)

- 54 CPMS case discussions since October 2017

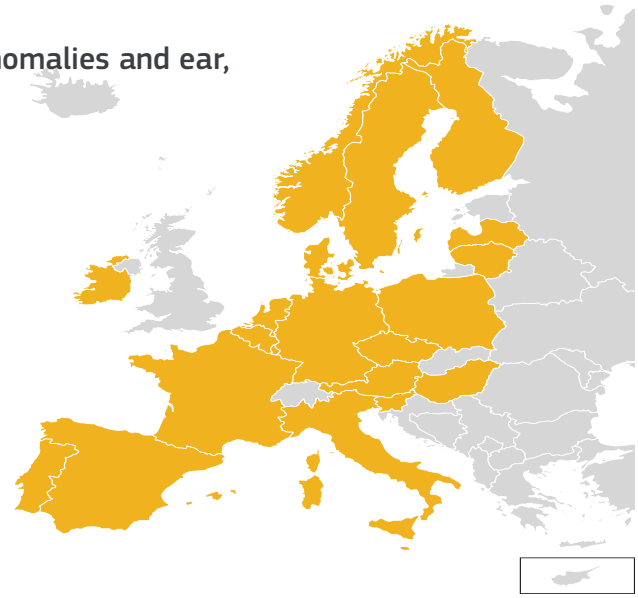
Research and patients' registries

- 32 relevant research projects or clinical trials involving at least two healthcare providers from two different Member States
- 4 223 patients (bone dysplasia - calcium and phosphate) recorded in the EuRR-Bone registries (https://eurreb.eu/), a shared platform with Endo-ERN

ERN CRANIO

European Reference Network on rare craniofacial anomalies and ear, nose and throat (ENT) disorders

https://www.ern-cranio.eu/



Members

42 healthcare providers located in 21 countries in the EU and Norway are members of the network, distributed as follows:

- 35 Full Members in 14 Member States, plus Norway
- 5 Associated National Centres in 4 Member States
- 2 National Coordination Hubs in 2 Member States

In addition, 9 patients' representatives and 7 supporting partners are collaborating

Disease areas

- Craniofacial anomalies
- Cleft lip/palate and odontological disorders
- ENT (Ear, Nose & Throat) disorders

Guidelines, care pathways, and patient journeys

- 5 guidelines written by ERN CRANIO
- 1 consensus statement written by ERN CRANIO
- 6 guidelines co-authored and endorsed by ERN CRANIO
- 2 patient-friendly guidelines
- 3 patient journeys

Training and education

- 12 webinars organised since 2017
- 60+ exchanges organised since 2017
- 1 course accredited by the Union Européenne des Médecins Spécialistes

Clinical Patient Management System (CPMS)

- 43 CPMS case discussions since 2017

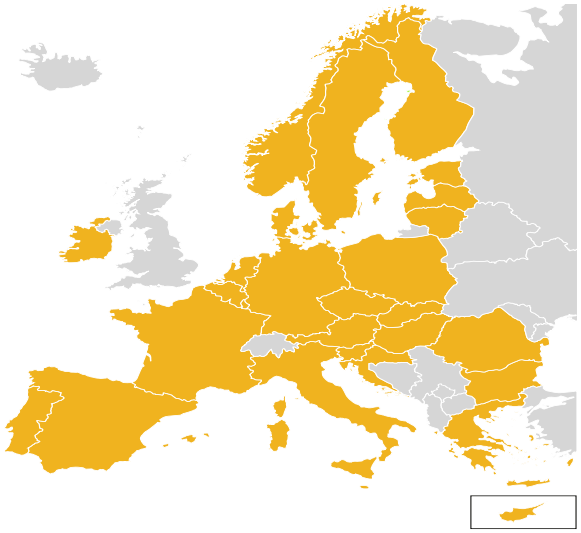
Research and patients' registries

- 45 relevant research projects or clinical trials involving at least two healthcare providers from two different Member States.
- 3 sub-registries developed and in use
- 6 sub-registries in development
- 900+ patients recorded in the ERN CRANIO registry (https://erncranio.molgenis.net/CranioPublic/cranio-public/#/)

ENDO ERN

European Reference Network on rare endocrine conditions

https://endo-ern.eu/



Members

105 healthcare providers located in 27 countries in the EU and Norway, distributed as follows:

- 91 Full Members in 21 Member States
- 11 Associated National Centres in 5 Member States and 2 in Norway
- 1 National Coordination Hub in a Member State

20 patient representatives from 10 countries are also collaborating.

Disease areas

- Adrenal
- Disorders of Calcium & Phosphate Homeostasis
- Genetic Disorders of Glucose & Insulin Homeostasis
- Genetic Endocrine Tumour Syndromes
- Growth & Genetic Obesity Syndromes
- Hypothalamic and Pituitary Conditions
- Sex Development & Maturation
- Thyroid

Guidelines, care pathways, and patient journeys

- 4 guidelines written by Endo-ERN
- About 60 guidelines co-authored and/or endorsed by Endo-ERN
- 1 care pathway
- 3 patient journeys

Training and education

- 70+ Endo-ERN webinars organised since 2019
- Joint webinar programme with European Endocrine societies (adult and paediatric)
- Endo-ERN symposium at European Endocrine Societies Annual Conferences
- 17 clinical exchanges (20 health care providers, 12 countries) since 2021
- Endorsement / accreditation of educational activities

Clinical Patient Management System (CPMS)

- 250 CPMS case discussions since 2017

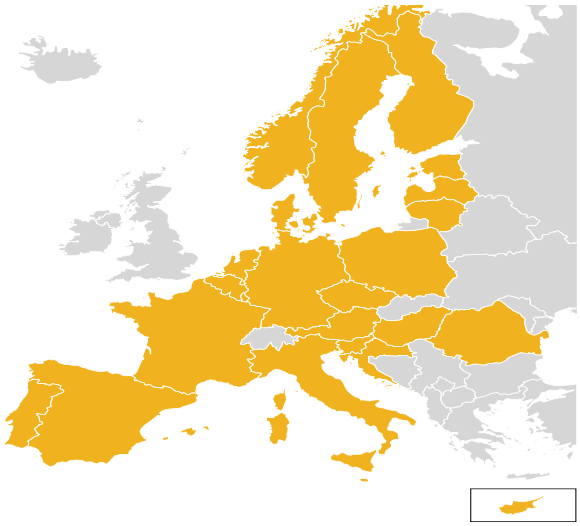
Research and patients' registries

- About 200 relevant research projects or clinical trials involving at least two healthcare providers from two different Member States
- 3 850 patients recorded in the core registry (https://eurreb.eu), a shared platform with ERN BOND

ERN EpiCARE

European Reference Network on rare and complex epilepsies

https://epi-care.eu/



Members

50 healthcare providers located in 24 countries in the EU and Norway are members of the network, distributed as follows:

- 38 Full Members in 15 Member States, plus Norway
- 10 Associated National Centres in 6 Member States
- 2 National Coordination Hubs in 2 Member States

In addition, 22 patient representatives (patient associations) are collaborating and collaborative agreements are in place with the 3 main scientific societies in the field (European section of the International League Against Epilepsy; European Academy of Neurology; European Paediatric Neurology Society).

Disease areas

- 160 rare forms of epilepsy, mostly of genetic origin.
- Highly complex cases of focal epilepsies, candidates to a pre-surgical evaluation and epilepsy-surgery

Guidelines, care pathways, and patient journeys

- 9 guidelines written by ERN EpiCARE
- 12 guidelines co-authored with the scientific societies and endorsed by ERN EpiCARE
- 20 patient leaflets in progress
- 20 patient journeys (9 published) and an emergency protocol tool

Training and education

- 85 webinars were held, with the participation of international experts, both from EpiCARE centres and elsewhere / 2 COVID-19 specific webinars were organised in response to the pandemic (2020) and one on the Ukraine war
- Exchange programme supporting nurses working in epilepsy units and neuropsychologists to exchange on local practices
- Support for young clinicians in presenting their work at the annual scientific workshop held in Rome (30 participations in 4 years) and to congresses of the scientific societies (10 clinicians)
- Mentor-mentee programme in progress; 30 young clinicians have participated in the Rome workshop over the past 4 years

Clinical Patient Management System (CPMS)

- 230 CPMS case discussions since 2017

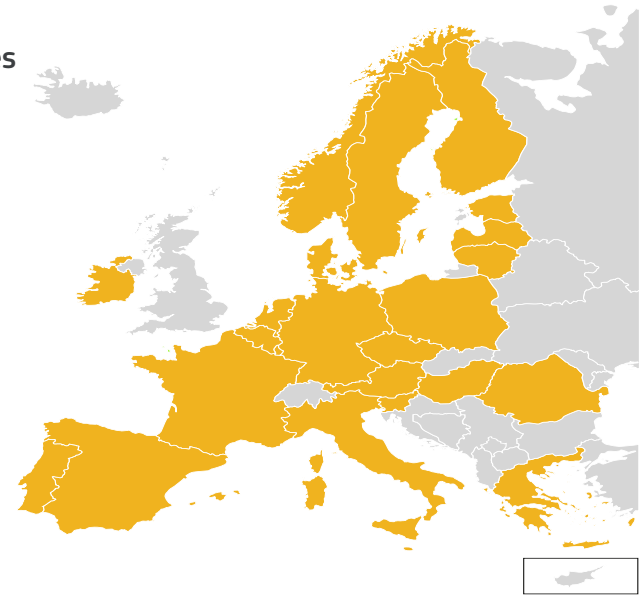
Research and patients' registries

- 85 research projects and clinical trials, each involving at least two healthcare providers from two different Member States that belong to EpiCARE
- at least 70% of all centres (35 of 50 member centres) are already using the EpiCARE registry REDCap template
- 9 500 patients are already registered in the local REDCap EpiCARE registry
- Fostering research Initiatives:
  - Genetic Collaborative Research Platform, promoting targeted research initiatives
  - A regularly updated Grant Opportunities platform

ERKNet

European Reference Network on rare kidney diseases

https://www.erknet.org/



Members

74 healthcare providers located in 24 countries in the EU and Norway are members of the network, distributed as follows:

- 64 Full Members in 18 Member States
- 7 Associated National Centres in 4 Member States and 1 in Norway
- 2 National Coordination Hubs in 2 Member States

Disease areas

- Glomerulopathies
- Congenital Malformations & Ciliopathies
- Tubulopathies
- Metabolic & Stone Disorders
- Thrombotic Microangiopathies
- Paediatric CKD & Dialysis
- Paediatric Transplantation
- Rare Causes of Hypertension

Guidelines, care pathways, and patient journeys

- 14 guidelines written by ERN ERKNet
- 62 guidelines endorsed by ERKNet
- 3 care pathways in progress
- 12 patient journeys developed

Training and education

- 121 webinars organised since February 2018
- 6 continuous medical education courses organised
- 15 research mobility exchanges
- 64 clinical exchanges (30 healthcare providers, 17 countries) since 2021
- A structured 3-year postgraduate curriculum focusing on rare kidney diseases integrating e-learning modules, interactive webinars, and hands-on clinical experience

Clinical Patient Management System (CPMS)

- 40 CPMS case discussions since May 2018

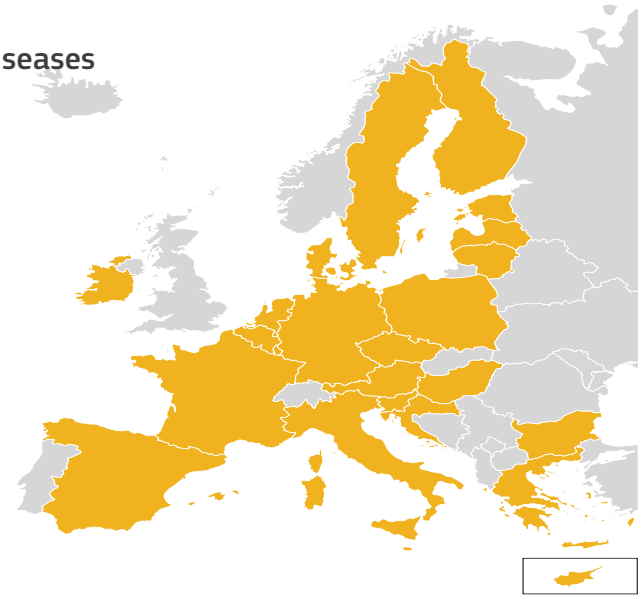
Research and patients' registries

- Participation in 56 research projects or clinical trials involving at least two healthcare providers from two different Member States
- >27 000 patients registered in the ERKReg registry (https://www.erknet.org/patients-registry/registry-mission).

ERN-RND

European Reference Network on rare neurological diseases

https://www.ern-rnd.eu/



Members

70 healthcare providers located in 24 EU countries are members of the network, distributed as follows:

- 63 Full Members in 20 Member States
- 2 Associated National Centres in 2 Member States
- 2 National Coordination Hubs in 2 Member States

In addition, collaborations with 9 patient representatives are in place

Disease areas

- Ataxia and Hereditary Spastic Paraplegias
- Huntington's Disease and Choreas
- Dystonia, paroxysmal disorders and Neurodegeneration with Bran Iron Accumulation
- Leukoencephalopathies
- Atypical parkinsonian syndromes
- Frontotemporal dementia

Guidelines, care pathways, and patient journeys

- 6 patient journeys in up to 15 different languages
- 10 adopted clinical practice guidelines
- 4 clinical practice guidelines in development
- 13 consented and implemented care pathways

Training and education

- European Training Requirement for Rare Neurological Diseases developed by ERN-RND adopted by European Union of Medical Specialists (September 2024)
- 100 webinars organised (as of August 2024)
- Short exchange program: until the end of 2023, 16 healthcare professionals visited 12 host institutions
- 5 winter and spring schools on highly specialised healthcare services such as neurorehabilitation (2022), neuroimaging (2023) and deep brain stimulation (2024)
- Postgraduate curriculum for Rare Neurological Diseases under development together with the European Academy of Neurology and the European Paediatric Neurology Society implementing the European Training Requirement for Rare Neurological Diseases

Clinical Patient Management System (CPMS)

- 323 CPMS case discussions since 2017

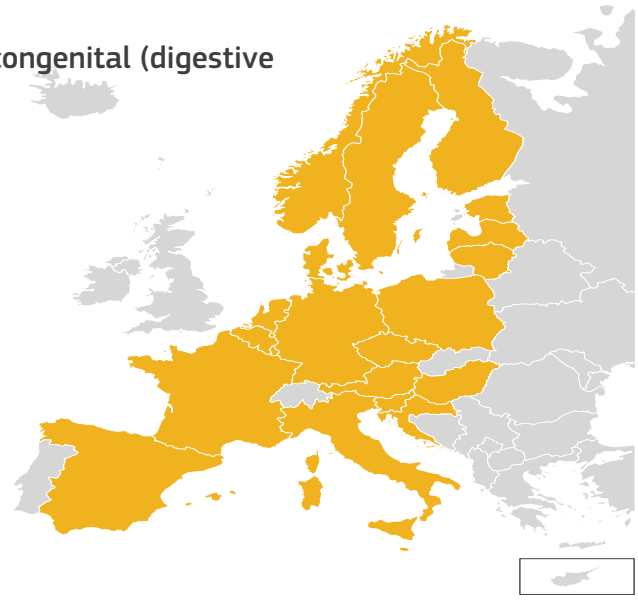
Research and patients' registries

- ERN-RND members participate in 12 observational studies and 16 clinical trials involving at least two healthcare providers from two different Member States.
- ERN-RND members have a leading role in flagship European rare diseases research projects and initiatives such as Solve-RD (www.solve-rd.eu) and ERDERA (www.erdera.org).
- > 12 000 patients in the registry in 2024 (https://www.ern-rnd.eu/ern-rnd-registry/?cn-reloaded=1#registry-objectives)

ERNICA

European Reference Network on rare inherited and congenital (digestive and gastrointestinal) anomalies

https://www.ern-ernica.eu/



Members

52 healthcare providers located in 21 countries in the EU and Norway are members of the network, distributed as follows:

- 39 Full Members in 11 Member States, plus Norway
- 9 Associated National Centres in 5 Member States
- 4 National Coordination Hubs in 4 Member States

In addition, collaborations with 13 patients' organisations and 4 individual parents of patients are in place

Disease areas

- Oesophageal diseases
- Intestinal diseases
- Gastroenterological diseases
- Intestinal failure
- Abdominal wall defects
- Malformations of the diaphragm

Guidelines, care pathways, and patient journeys

- 6 guidelines written by ERNICA
- 2 patient journeys

Training and education activities

- 44 webinars organised in 2020-2023
- 1/2 Colorectal hands-on training courses supported by ERNICA per year and one Hands-on Training Course Congenital Diaphragmatic Hernia and extracorporeal membrane oxygenation (ECMO) per year (previously supported and now fully organised by ERNICA), one course on prenatal assessment, and development of a Flagship Surgical Training Programme in cooperation with eUROGEN.
- 2 clinical exchange programmes with 15 visitors in 2023
- 52 developed/endorsed educational videos and animations on YouTube

Clinical Patient Management System (CPMS)

- 5 CPMS case discussions

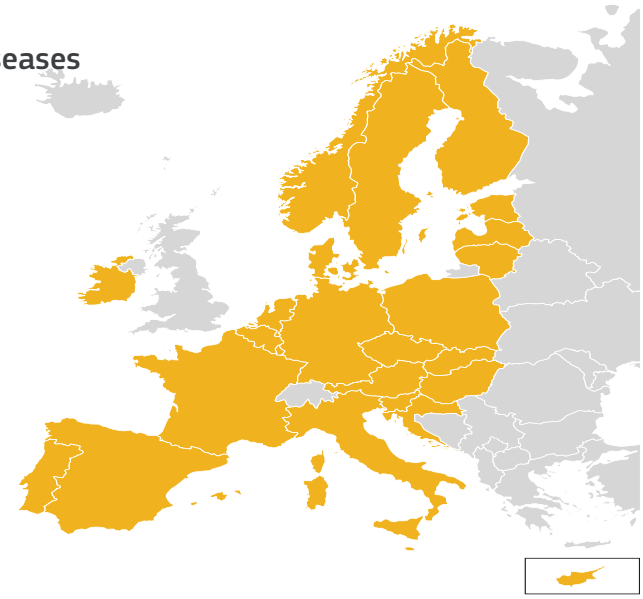
Research and patients' registries

- 24 relevant research projects or clinical trials involving at least two healthcare providers from two different Member States in 2023 (more related project are listed in the ERNICA Research Catalogue: https://www.ern-ernica.eu/ernica-research-catalogue)
- 30 centres connected to the EPSA|ERNICA registry (https://www.ern-ernica.eu/registry) in 2023, including 3 076 patients

ERN-LUNG

European Reference Network on rare respiratory diseases

https://ern-lung.eu



Members

87 healthcare providers located in 25 countries in the EU and Norway are members of the network, distributed as follows:

- 78 Full Members in 18 Member States, plus Norway
- 7 Associated National Centres in 4 Member States
- 2 National Coordination Hubs in 2 Member States

Disease areas

- Alpha-1 antitrypsin deficiency (AATD)
- Bronchiectasis (BE)
- Chronic Lung Allograft Dysfunction (CLAD)
- Cystic Fibrosis (CF)
- Interstitial Lung Diseases (ILD)
- Mesothelioma (MSTO)
- Other Rare Lung Diseases (ORLD)
- Primary Ciliary Dyskinesis (PCD)
- Pulmonary Hypertension (PH)
- Sarcoidosis (SARC)

Guidelines, care pathways, and patient journeys

- 6 guidelines endorsed by ERN LUNG
- 109 publications by ERN-LUNG members
- Patient journeys for 4 ERN-LUNG disease areas
- Patient Priorities project for 3 core networks (Sarcoidosis, Interstitial Lung Diseases, and Bronchiectasis

Training and education

- ERN-LUNG Academy launched in 2023. Participants watch webinars and make a 1-week practical stay in one of the 79 healthcare providers, fully reimbursed (for EU clinicians). For 2024, 41 participants from 12 EU Member States and 5 non-EU Member States. The webinars were produced by the network's clinicians
- ERS/ERN-LUNG Virtual School of Rare Lung Diseases: this yearly online conference, which launched in 2023 and ran for the second time in 2024, had over 100 participants

Clinical Patient Management System (CPMS)

- 68 CPMS case discussions since 27 November 2024

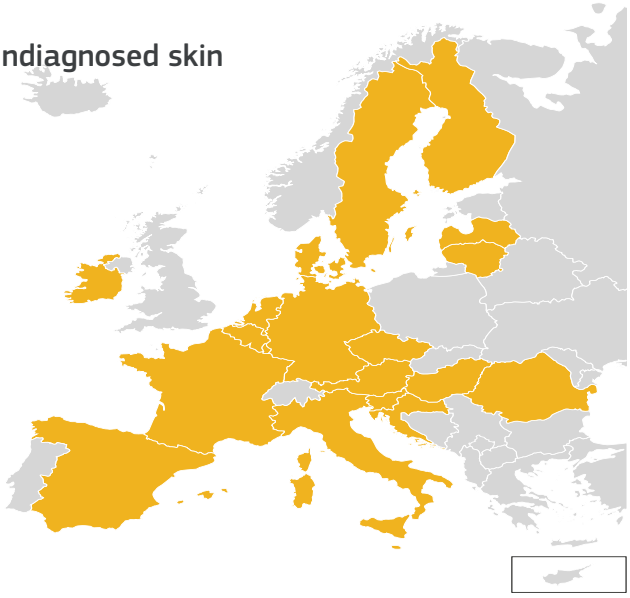
Research and patients' registries

- 9 Research projects were supported, and 3 three Core networks (cystic fibrosis, Alpha-1 antitrypsin deficiency, Primary Ciliary Dyskinesis) have each implemented a well-established clinical trial network. Others, like Bronchiectasis, and Pulmonary Hypertension, are in their early phase
- About 2 000 patient data are recorded in the ERN-LUNG PRIME (https://ern-lung.eu/patient-registry/ern-lung-registry/)
- 160 patients have been recorded in the population registry BREATHeREGISTRY (https://ern-lung.eu/patient-registry/population-registry/) as of July 2024

ERN-Skin

European Reference Network on rare, complex, and undiagnosed skin disorders

https://ern-skin.eu/



Members

56 healthcare providers located in 20 EU countries are members of the network, distributed as follows:

- 52 Full Members in 16 Member States
- 2 Associated National Centres in 2 Member States
- 2 National Coordination Hubs in 2 Member States

In addition, collaborations with 16 patient representatives are in place

Disease areas

- Inherited epidermolysis bullosa and skin fragility syndromes, Darier, Hailey-Hailey
- Ichthyosis and palmoplantar keratoderma
- Ectodermal dysplasias including incontinentia pigmenti and p63-associated disorders
- Mendelian causes of connective tissue disorders
- Cutaneous mosaic disorders - nevi and nevoid skin disorders and complex vascular malformations and vascular tumours
- Cutaneous diseases related to DNA repair disorders and photosensitivity
- Autoimmune bullous diseases severe cutaneous drug reactions
- Hidradenitis suppurativa - PAPA, PAPASH, PASH, PASS, SAPHO - Behçet, Degos

Guidelines, care pathways, and patient journeys

- 18 guidelines written by ERN-Skin
- 28 guidelines co-authored and endorsed by ERN-Skin
- 8 patient journeys

Training and education

- 29 webinars organised since 2021
- 52 short exchange programmes
- 1 ERN-Skin e-training platform accredited by the UEMS (European Union of Medical Specialists)
- World Congress on Rare Skin Disease 2022 & 2024

Clinical Patient Management System (CPMS)

- 167 CPMS case discussions since 2019

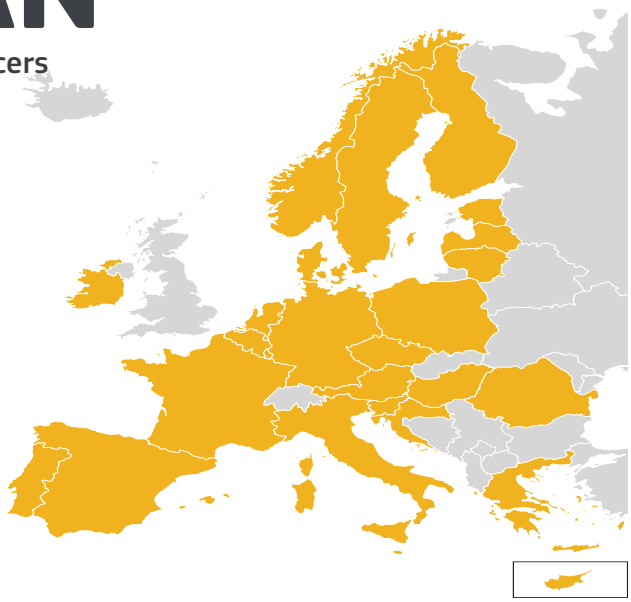
Research and patients' registries

- 78 relevant research projects or clinical trials involving at least two healthcare providers from two different Member States
- 11 patients recorded in the ERN-Skin registry (https://ern-skin.eu/erras-registry/)

ERN EURACAN

European Reference Network on rare adult solid cancers

https://euracan.eu/



Members

101 healthcare providers located in 26 countries in the EU and Norway are members of the network, distributed as follows:

- 92 Full Members in 18 Member States, plus Norway
- 7 Associated National Centres in 5 Member States
- 2 Coordination Hubs in 2 Member States

14 patients' representatives are also collaborating

Disease areas

- Connective tissue (sarcomas)
- Female genital organs and placenta
- Male genital organs and urinary tract
- Neuroendocrine system
- Digestive tract
- Endocrine organs
- Head and neck
- Thorax
- Skin and eye melanoma
- Brain and spinal cord

Guidelines, care pathways, and patient journeys

- 18 guidelines co-authored with scientific societies and 6 endorsed by ERN EURACAN

Training and education

- 22 online courses organised by the ERN EURACAN + 42 lectures organised in collaboration with the European School of Oncology since 2017
- Short exchange programme: 40 participants from around 30 healthcare providers since 2021
- Professorship programme since 2023

Clinical Patient Management System (CPMS)

- 106 CPMS case discussions since January 2022

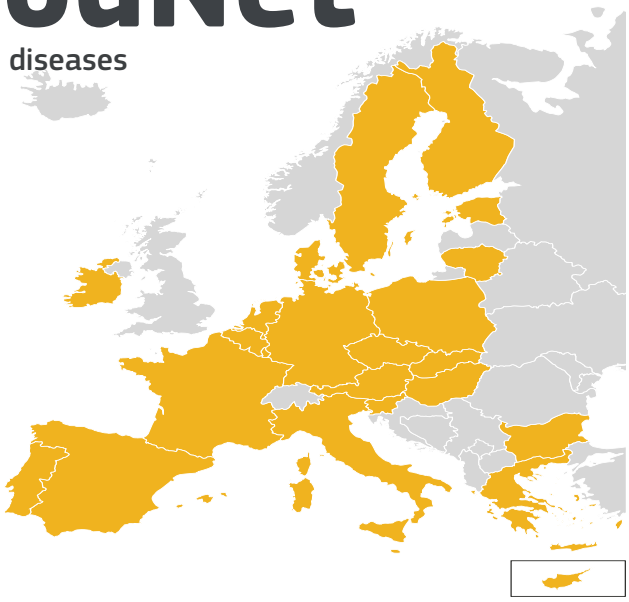
Research and patients' registries

- 844 relevant research projects or clinical trials involving at least two health care providers from two different Member States
- 6 600 patients included in the registries. At the end of 2023, the head and neck cancer registry (period of diagnosis 2018-2022) had 600 cases. The sarcoma registry currently combines 2 data bases with 6 000 soft tissue sarcoma cases (period of diagnosis 2000 onward)

ERN EuroBloodNet

European Reference Network on rare haematological diseases

https://eurobloodnet.eu/



Members

97 healthcare providers located in 24 EU countries are members of the network, distributed as follows:

- 90 Full Members in 18 Member States
- 4 Associated National Centres in 3 Member States
- 3 National Coordination Hubs in 3 Member States

In addition, collaborations with 11 European Patient Advocacy Groups (ePAGs) and 52 national patient organisations involved in ERN actions are in place

Disease areas

4 non-oncological disease areas (paediatrics and adults):

- Rare Red blood cell defects
- Bone marrow failure and hematopoietic disorders
- Rare bleeding-coagulation disorders and related diseases
- Hemochromatosis and other rare genetic disorders of iron metabolism and heme synthesis

2 oncological disease areas (adults):

- Lymphoid malignancies
- Myeloid malignancies

Guidelines, care pathways, and patient journeys

- 4 guidelines written, 4 under development
- 70 guidelines endorsed by ERN-EuroBloodNet
- 7 EU mappings on availability of Highly Specialised Procedures (diagnosis/prevention/treatment) standards of care not available in all EU, of which two published so far, i.e: Diagnostic procedures for Primary vitreoretinal lymphoma in Europe, Transcranial Doppler screening and stroke prevention for children with sickle cell disease in Europe.
- 1 patient journey under development

Training and education

- 330 educational trainings (including webinars, videos and patients' onsite trainings) organised since 2019
- 13 short exchange programmes organised since 2019
- 8 training courses endorsed since 2022
- 1 publication on ERN-EuroBloodNet Educational Strategy

Clinical Patient Management System (CPMS)

- 57 CPMS case discussions since 2017

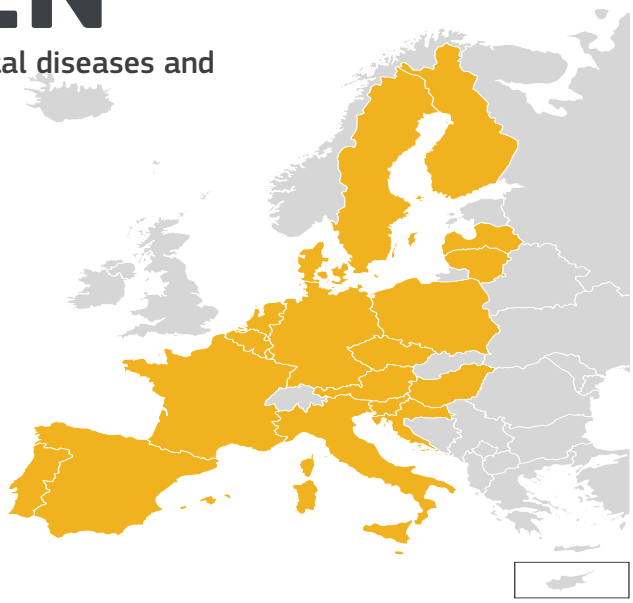
Research and patients' registries

- 95 clinical trials involving at least two healthcare providers from two different Member States in 2023, 1 of them acknowledging the ERN
- The first trial (SATISFY) has completed accrual, and the second (LUSPARA) is about to be submitted to the European Clinical Trials Information System for clinical trials (CTIS)
- 18 observational studies involving at least two healthcare providers from two different member states in 2023, 11 of them acknowledging the ERN
- Leaders of work package 3 in ERICA (European Rare Disease Research Coordination and Support Action) and of work package 10 in ERDERA (European Rare Disease Research Alliance)
- 4 685 new patients transferred to ENROL registry (www.enrolnetwork.eu) during 2024

ERN eUROGEN

European Reference Network on rare uro-recto-genital diseases and complex conditions

https://eurogen-ern.eu/



Members

56 healthcare providers located in 20 EU countries are members of the network, distributed as follows:

- 51 Full Members in 15 Member States
- 1 Associated National Centre in 1 Member State
- 4 Coordination Hubs in 4 Member States

7 patient representatives are also collaborating

Disease areas

Three Work Streams (WS):

- WS 1: rare congenital uro-recto-genital anomalies (paediatrics).
- WS 2: functional uro-recto-genital conditions requiring highly specialised surgery (adults).
- WS 3: rare uro-recto-genital tumours.

Each work stream is divided into expertise areas containing the rare diseases and complex conditions that are covered.

Guidelines, care pathways, and patient journeys

- 3 new Clinical Practice Guidelines written (including one in collaboration with ERN ITHACA and ERN ERKNet), 2 more under development
- 11 Clinical Decision Support Tools under development
- 1 Care Pathway under development
- 5 Patient Journeys, plus other patient information

Training and education

- 113 webinars organised since June 2019
- 1 short exchange programme (1 February to 31 August 2023)
- 4 other training activities (3 surgical colorectal courses + 1 flagship surgical training programme in development with ERN ERNICA)
- 1 book, “Rare and Complex Urology”, written by network experts and published by Elsevier

Clinical Patient Management System (CPMS)

- 205 CPMS case discussions since June 2017

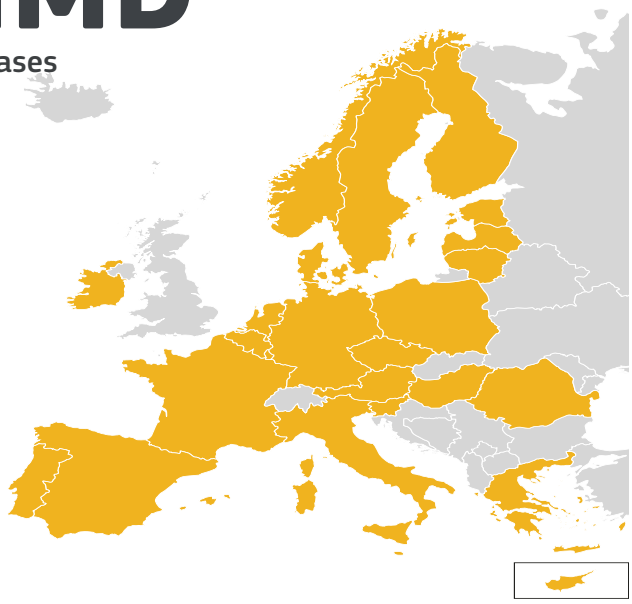
Research and patients’ registries

- 7 research projects or clinical trials involving at least two healthcare providers from two different Member States
- 1 011 patients registered in the ERN eUROGEN registry (https://eurogen-ern.eu/our-work/registry/)

ERN EURO-NMD

European Reference Network on neuromuscular diseases

https://ern-euro-nmd.eu/



Members

82 healthcare providers located in 25 countries in the EU and Norway are members of the network, distributed as follows:

- 74 Full Members in 17 Member States, plus Norway
- 6 Associated National Centres in 5 Member States
- 2 National Coordination Hubs in 2 Member States

In addition, 34 patients 'representatives from 28 patient organisations and over 15 countries are collaborating

Disease areas

- Muscle Diseases
- Neuromuscular Junction disorders
- Motor neuron diseases
- Peripheral Nerve disorders
- Mitochondrial diseases

Guidelines, care pathways, and patient journeys

- 6 guidelines written by ERN EURO-NMD
- 26 guidelines co-authored and 26 endorsed by ERN EURO-NMD
- 3 patient journeys on myasthenia gravis, myofibrillar myopathy and multifocal motor neuropathy (3 more in progress)

Training and education

- 93 webinars held since 2019, including a webinar series on gene therapy, mitochondrial diseases, peripheral nerve diseases, and neuromuscular pathology
- 23 training exchanges at 8 host sites across 6 countries (154 total training days)
- Every year since 2018, a summer school on translational research has been held in collaboration with TREAT-NMD, a global network of experts in the neuromuscular field (7 editions to date) and, since 2023, the summer school on multidisciplinary care (2 editions to date)
- Moodle-based EURO-NMD Academy, offering a certified six-module course on mitochondrial disorders (efforts underway for accreditation with the European Union of Medical Specialists - UEMS)

Clinical Patient Management System (CPMS)

- 140 CPMS case discussions since 2017

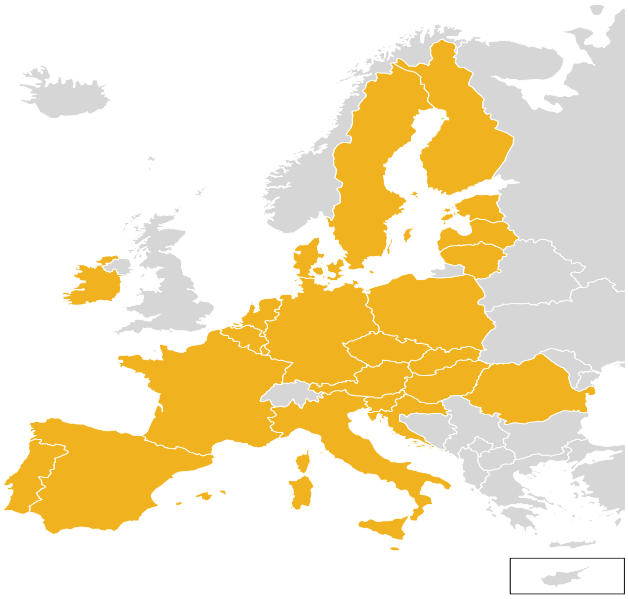
Research and patients' registries

- 165 relevant research projects or clinical trials involving at least two health care providers from two different Member States
- 25 patients enrolled in the EURO-NMD Registry (https://registry.ern-euro-nmd.eu/) since its launch in January 2024

ERN-EYE

European Reference Network on rare eye diseases

https://www.ern-eye.eu/



Members

59 healthcare providers located in 24 EU countries are members of the network, distributed as follows:

- 51 Full Members in 18 Member States
- 5 Associated National Centres in 3 Member States
- 3 National Coordination Hubs in 3 Member States

In addition, collaborations with 9 patients 'representatives are in place

Disease areas

- Retinal Diseases
- Neuro ophthalmology
- Paediatric Rare Eye Diseases
- Anterior Segment

Guidelines, care pathways, and patient journeys

- 1 consensus statement written by ERN-EYE ERNKnet, Endo-ERN, and ERN-ITHACA
- 6 consensus statements being finalised

Training and education

- 10 webinars organised since 2021
- 3 short exchange programmes
- 1 eLearning programme on Inherited Retinal Diseases accredited by the European Accreditation Council for Continuing Medical Education (EACCME)
- Educational videos
- 1 Serious game on how to announce a diagnosis

Clinical Patient Management System (CPMS)

- 163 CPMS case discussions since 2017

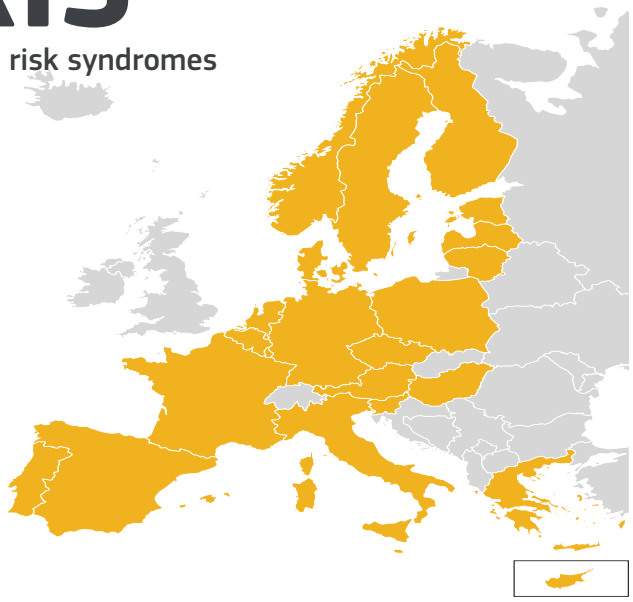
Research and patients' registries

- 31 relevant research projects and clinical trials involving at least two healthcare providers from two different Member States
- 6 patients included in the ERN-EYE registry (https://redgistry.eu/)

ERN GENTURIS

European Reference Network on rare genetic tumour risk syndromes

https://www.genturis.eu/l=eng/home.html



Members

51 healthcare providers located in 23 countries in the EU and Norway are members of the network, distributed as follows:

- 44 Full Members in 17 Member States
- 4 Associated National Centres in 3 Member States and 1 in Norway
- 2 National Coordination Hubs in 2 Member States

Disease areas

- Schwannomatosis & neurofibromatosis
- Lynch syndrome and polyposis
- Hereditary breast and ovarian cancer syndrome
- Other rare – predominantly malignant

Guidelines, care pathways, and patient journeys

- 6 guidelines written by ERN GENTURIS
- 3 guidelines co-authored and 17 endorsed by ERN GENTURIS
- Care pathways for all ERN GENTURIS disease areas
- Patient journeys for all ERN GENTURIS disease areas

Training and education

- ERN GENTURIS e-Training Programme: about 60 free webinars available on demand
- Every even year, 1 course in Italy (Bertinoro) for geneticists on Hereditary Cancer Genetics
- Every odd year, 1 course in France (Paris) for medical oncologists on Hereditary Cancer Genetics

Clinical Patient Management System (CPMS)

- 285 CPMS case discussions since 2017

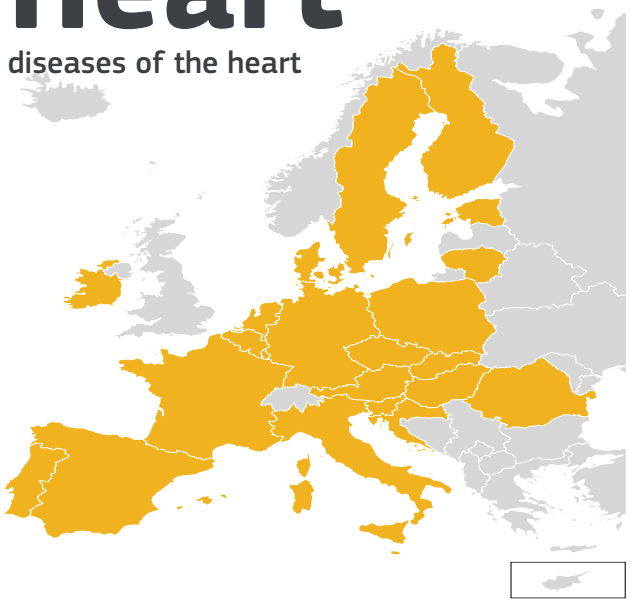
Research and patients' registries

- 12 research projects in which at least two ERN GENTURIS healthcare providers from different countries are participating
- 1 600+ patients in the ERN GENTURIS Registry (https://genturis-registry.eu/menu/main/app-ern-genturis/)

ERN GUARD-Heart

European Reference Network on uncommon and rare diseases of the heart

https://guardheart.ern-net.eu/



Members

52 healthcare providers located in 23 EU countries are members of the network, distributed as follows:

- 43 Full Members in 16 Member States
- 7 Associated National Centres in 5 Member States
- 2 National Coordination Hubs in 2 Member States

In addition, collaborations with 23 patient representatives (15 European Patient Advocacy Groups and 8 supporting partners) are in place

Disease areas

- Familial electrical diseases
- Familial cardiomyopathies
- Special electrophysiology conditions in children
- Congenital heart diseases
- Other rare heart diseases

Guidelines, care pathways, and patient journeys

- 24 guidelines (co)-authored and endorsed by ERN GUARD-Heart
- 1 patient journey

Training and education

- 24 webinars organised by since July 2022
- ongoing exchange programme (15 packages per ERN per year)
- 22 layperson abstracts of ERN publications available on website
- 1 summer school programme per year

Clinical Patient Management System (CPMS)

- 173 CPMS case discussions since 2018

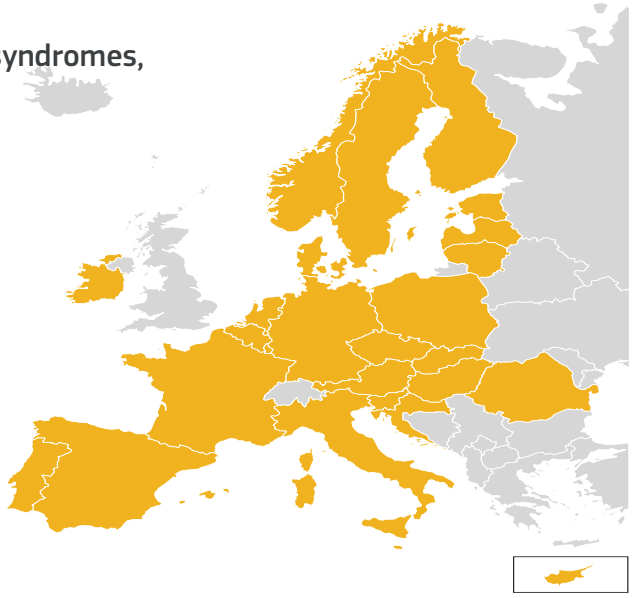
Research and patients' registries

- 119 relevant peer reviewed publications involving at least two healthcare providers from two different member states.
- 12 disease-specific registries, and another 11 in preparation (https://guardheart.ern-net.eu/disease-registries/)
- > 8 500 patients recorded in the Heart-Core registry (https://guardheart.ern-net.eu/disease-registries/heart-core-registry/)

ERN-ITHACA

European Reference Network on rare malformation syndromes, intellectual and other neurodevelopmental disorders

https://ern-ithaca.eu/



Members

71 healthcare providers located in 26 countries in the EU and Norway are members of the network, distributed as follows:

- 66 Full Members in 21 Member States
- 1 Associated National Centre in 1 Member State and 1 in Norway
- 3 National Coordination Hubs in 3 Member States

The Patient Advisory Board represents over 60 national or European Patient Advocacy Groups

Disease areas

ITHACA covers both developmental anomalies (DA) and neurodevelopmental disorders (NDD) of genetic, genomic/chromosomal or environmental origin, and those resulting from teratogen exposure.

1.Disorders of human development (over 5 000 Orphanet entries)

- Congenital malformations (single/multiple) and dysmorphisms
- Spina bifida and related dysraphisms (shared area with ERN eUROGEN)
- Foetal medicine
  - Prenatal diagnosis of foetal anomalies
  - Foetal surgery for spinal bifida
  - Foetal pathology
  - Disorders of monozygotic twinning

2.Disorders of human neurodevelopment (over 2 500 genes)

- Intellectual disability
- Autism spectrum disorder
- Psychiatric manifestations of developmental disorders

Guidelines, care pathways, and patient journeys

- 5 clinical practice guidelines achieved and several others in their final stage): disorder-specific or transversal
- Contribution to 1 clinical practice guideline developed with ERN-EYE
- 5 patient journeys

Training and education

- EuroNDD: bi-annual multidisciplinary 2-day workshop on neurodevelopmental disorders (NDD)
- EuroDysmorpho: 4-day workshop, annually
- Multidisciplinary Foetal Diagnostics Winter School, annually
- ITHACA e-Training Programme: 14 free webinars on demand. New webinars are organised monthly.
- APOGeE (A Practical Online Genetics e-Education): a free online handbook on medical genetics
- MOOC (Massive open online course) “BIG” (bioinformatics in genetics)
- MOOC “Diagnosing rare diseases: from the Clinic to Research and back.”
- Writing/editing > 80 clinical summaries for ORPHANET
- Contribution to Orphanet classification update and enrichment of Human Phenotype Ontology thesaurus

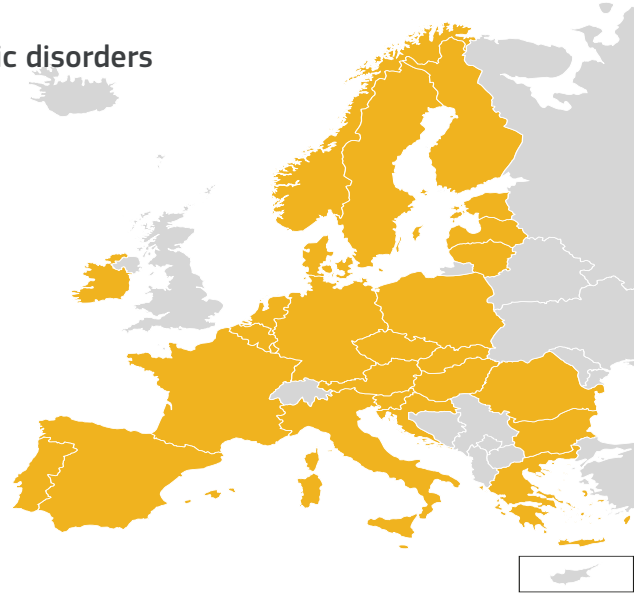
Research and patients’ registries

- Over 210 calls for collaborative call for collaboration in clinical research
- ILIAD: mixed (central + federative) registry, developed as a Health Data Warehouse (https://ern-ithaca.eu/our-research-activities/iliad/) of patients with NDD and/or DA. Collects the JRC minimal dataset and an ERN-specific minimal genetic dataset (genic and genomic/chromosomal variant). ILIAD offers the possibility of developing detailed sub-registries. Currently in beta phase (publicly accessible T2 2025). ILIAD registers three types of patients:
  - genetically defined patients (patients must have a genetic/genomic diagnosis to be recorded),
  - clinically defined patients (patients must have a precise clinical diagnosis with a disease-level ORPHA code),
  - patients with unknown etiopathological diagnosis and recognisable phenotype

MetabERN

European Reference Network on hereditary metabolic disorders

https://metab.ern-net.eu/



Members

91 healthcare providers located in 27 countries in the EU and Norway are members of the network, distributed as follows:

- 85 Full Members in 23 Member States, plus Norway
- 4 Associated National Centres in 1 Member State
- 2 National Coordination Hubs in 2 Member States

Disease areas

- AOA - Amino and Organic acids-related disorders
- C-FAO - Carbohydrate, fatty acid oxidation and ketone bodies disorders
- CDG - Congenital Disorders of Glycosylation and disorders of intracellular trafficking
- LSD - Lysosomal Storage disorders
- NOMS - Disorders of neuromodulators and other small molecule
- PD - Peroxisomal disorders
- PM-MD -Pyruvate metabolism mitochondrial oxidative phosphorylation disorders, Krebs cycle defects, disorders of thiamine transport & metabolism

Guidelines, care pathways, and patient journeys

- Review/development of Care Pathways and Patient Journeys
- Over 16 new guidelines developed by MetabERN
- Participation in the production of White papers and Recommendations
- Improvement of Regulatory process for medicines

Training and education

- Participation and provision of input and insights from patients and healthcare providers in the education of patients
- More than 500 training activities hosted by member centres to improve the knowledge and training of healthcare professionals in the field of inherited metabolic disorders
- More than 500 enrolled learners in the implementation and promotion of the MetabERN Diagnostic Clinical and Therapeutic Education Programme

Clinical Patient Management System (CPMS)

- 141 CPMS case discussions since 2017

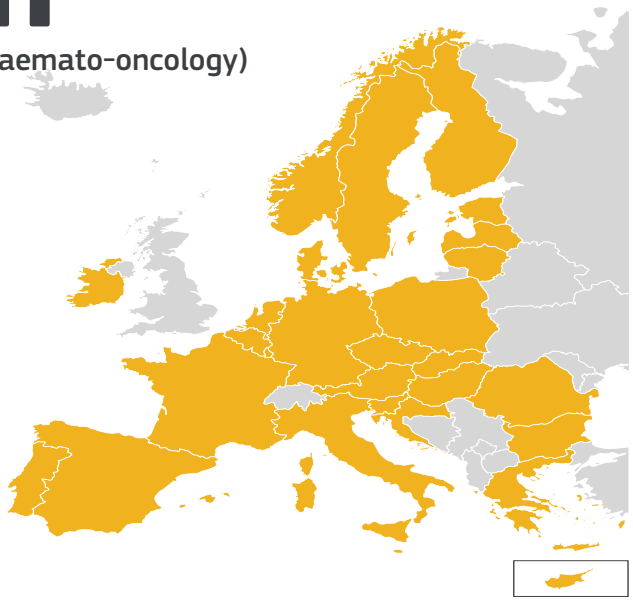
Research and patients' registries

- About 260 scientific publications
- Use of the U-IMD Registry (Unified European Registry for Inherited Metabolic Disorders, https://www.u-imd-registry.org/index.php?id=about): 30 participating centres, 3 534 registered patients
- Facilitation and harmonisation of new-born screenings in the EU: addition of a new module on the U-IMD Registry
- Creation of large databases with patients' data

ERN PaedCan

European Reference Network on paediatric cancer (haemato-oncology)

https://paedcan.ern-net.eu/



Members

90 healthcare providers located in 28 countries in the EU and Norway are members of the network, distributed as follows:

- 79 Full Members in 20 Member States, plus Norway
- 9 Associated National Centres in 5 Member States
- 2 National Coordination Hubs in 2 Member States

In addition, a collaboration with the largest pan-European childhood cancer parents´ survivors´ organisation, representing 64 member organisations in 35 countries, is in place

Disease areas

- Leukaemia
- Brain Tumours
- Lymphomas
- Neuroblastoma
- Renal Tumours
- Soft Tissue Sarcomas
- Bone Sarcomas
- Liver Tumours
- Germ Cell Tumours
- Retinoblastoma
- Very Rare Tumours (paediatric and adolescent and young adult population)

Guidelines, care pathways, and patient journeys

- 25 European Standard Clinical Practice guidelines written by ERN PaedCan

Training and education

- 42 webinars organised since 2021
- ERN PaedCan Training/Twinning programmes
- SIOP Europe (The International Society of Paediatric Oncology) Course in Paediatric Oncology
- SIOP Europe Virtual Courses
- SIOP Europe Student Summer School
- ESO (European School of Oncology)-SIOP Europe Masterclass, e-Learning, fellowships, and multidisciplinary course

Clinical Patient Management System (CPMS)

- 287 CPMS case discussions since 2018

Research and patients' registries

- 29 patients registered in the PARTNER registry for Very Rare Tumours (https://partner.datariverweb.com/myhealthtest/)
- European Standard of Clinical Practice (ESCP) registry on the delivery of standard clinical practice treatments across Europe. The ESCP registry has recently received a positive vote from the local ethics commission of the Medical University of Vienna for Austria. As a next step, the rollout strategy for other member states is being planned with the ERN PaedCan member healthcare providers.

ERN RARE-LIVER

European Reference Network on rare hepatological diseases

https://rare-liver.eu

Members

62 healthcare providers located in 23 countries in the EU and Norway are members of the network, distributed as follows:

- 52 Full Members in 15 Member States
- 6 Associated National Centres in 4 Member States and 1 in Norway
- 3 National Coordination Hubs in 3 Member States

In addition, collaborations with 15 patient representatives are in place

Disease areas

- Autoimmune liver diseases
- Metabolic, biliary atresia and related disease
- Structural liver disease

Guidelines, care pathways, and patient journeys

- 8 guidelines co-authored and 7 endorsed by ERN RARE-LIVER since 2017
- 3 care pathways (in progress)

Training and education

- 35 webinars organised since 2020
- Annual ERN RARE-LIVER ACADEMY (2-day onsite program)
- Variety of videos targeting actual topics in rare liver diseases

Clinical Patient Management System (CPMS)

- 526 CPMS case discussions since 2017

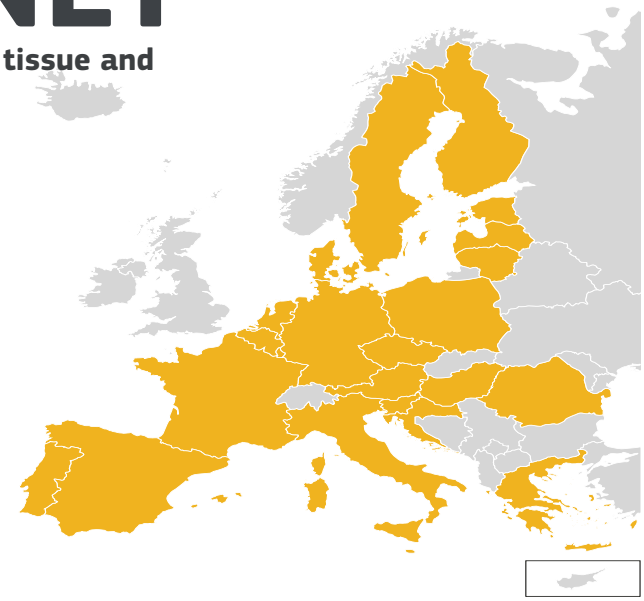
Research and patients' registries

- 6 relevant research projects and more than 50 clinical trials involving at least two health care providers from two different member states are currently ongoing
- 2 787 patients in the registry (https://rare-liver.eu/patients/r-liver-registry/)

ERN ReCONNET

European Reference Network on rare connective tissue and musculoskeletal diseases

https://reconnet.ern-net.eu/



Members

63 healthcare providers located in 23 EU countries are members of the network, distributed as follows:

- 54 Full Members in 15 Member States
- 6 Associated National Centres in 5 Member States
- 3 National Coordination Hubs in 3 Member States

In addition, collaborations with 17 patient representatives are in place

Disease areas

- Antiphospholipid Syndrome
- Ehlers-Danlos Syndromes
- Idiopathic Inflammatory Myopathies
- IgG4-Related Diseases
- Mixed Connective Tissue Diseases
- Relapsing Polychondritis
- Sjögren' Syndrome
- Systemic Lupus Erythematosus
- Systemic Sclerosis
- Undifferentiated Connective Tissue Diseases

Guidelines, care pathways, and patient journeys

- 12 scientific publications in the ERN ReCONNET Supplement "State of the Art of existing guidelines and unmet needs"
- 1 scientific publication reporting on the adherence, knowledge, and awareness of clinical practice guidelines in rare and complex connective tissue diseases
- 10 organisational care patient's pathway models co-designed with stakeholders applying the RarERN Path® - Methodology developed by ERN ReCONNET
- 1 scientific publication on the organisational care patient's pathway model developed on Systemic Sclerosis applying the RarERN Path® - Methodology developed by ERN ReCONNET
- 1 Red Flags on early diagnosis and referral published, 2 additional Red Flags already ongoing
- Points to consider for treating patients living with autoimmune rheumatic diseases with antiviral therapies and anti-SARS-CoV-2 antibody products
- 1 Set of Quality measures in transition of care in Rare and Complex Connective Tissue and Musculoskeletal Diseases (rCTDs)
- 1 lay version co-authored with patient representatives and published on the ERN ReCONNET website

Training and education

- 72 webinars organised by the ERN since 2019
- 43 short exchange programme visits organised so far
- Accredited online course to be launched in 2025
- Accredited course on transition of care

Clinical Patient Management System (CPMS)

- Over 50 CPMS case discussions since 2018

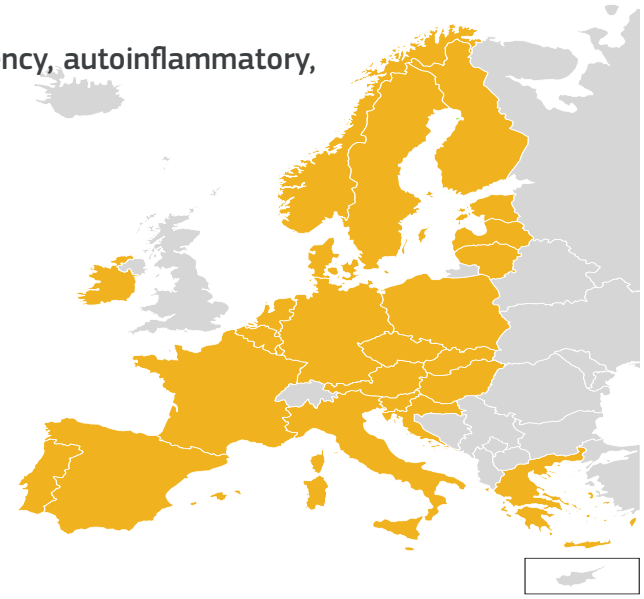
Research and patients' registries

- 7 clinical trials involving at least two health care providers from two different Member States
- Supplement "Rare inside Rare" to Clinical and Experimental Rheumatology, 2022, containing 18 scientific publications
- TogetherERN ReCONNET Registry Platform being finalised (https://reconnet.ern-net.eu/our-activities-registry/)
- VACCINATE – ERN ReCONNET multicentre prospective cohort study on vaccination
- Dedicated WG on Research and Quality of Care and WG on Registries and eHealth (https://reconnet.ern-net.eu/working-groups/)

ERN RITA

European Reference Network on rare immunodeficiency, autoinflammatory, and autoimmune and paediatric rheumatic diseases

https://ern-rita.org/



Members

70 healthcare providers located in 25 countries in the EU and Norway are members of the network, distributed as follows:

- 61 Full Members in 18 Member States, plus Norway
- 7 Associated National Centres in 4 Member States
- 2 National Coordination Hubs in 2 Member States

In addition, collaborations with 12 patient representatives are in place

Disease areas

- Primary immunodeficiencies
- Autoinflammatory disorders
- Autoimmune diseases
- Paediatric rheumatic diseases

Guidelines, care pathways, and patient journeys

- 8 guidelines co-authored and 3 endorsed by ERN RITA
- ERN RITA Patient Journey Handbook
- 3 Patient Journeys

Training and education

- 40 webinars organised since 2020
- Tuesday Lunch webinar series
- Patient-centred webinars
- Short exchange programmes among ERN RITA healthcare providers

Clinical Patient Management System (CPMS)

- 149 CPMS case discussions since 2017

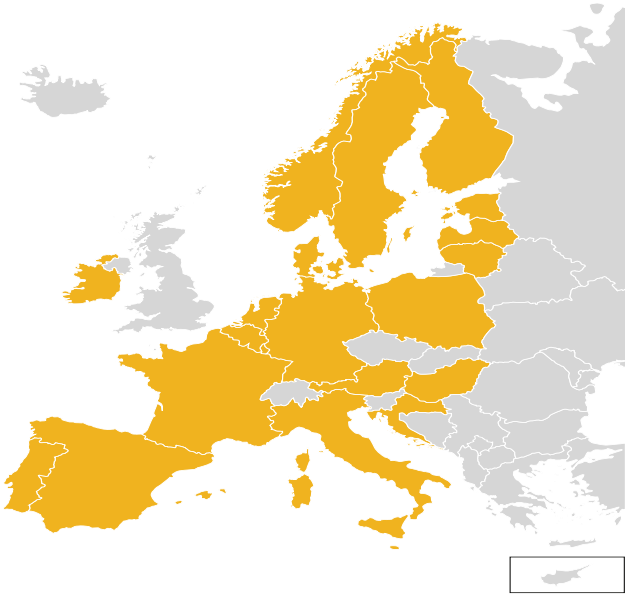
Research and patients' registries

- 126 relevant research projects or clinical trials involving at least two healthcare providers from two different Member States
- Development of a network registry that includes new patients treated by ERN RITA healthcare providers (https://ern-rita.org/rita-registry/general-info/)

ERN TRANSPLANT-CHILD

European Reference Network on transplantation in children

https://transplantchild.eu/



Members

40 healthcare providers located in 21 countries in the EU and Norway are members of the network, distributed as follows:

- 33 Full Members in 13 Member States, plus Norway
- 4 Associated National Centres in 4 Member States
- 3 National Coordination Hubs in 3 Member States

In addition, collaborations with 13 patient representatives are in place

Transplantation areas

- Solid organ transplantation (heart, lung, kidney, liver and intestinal)
- Haematopoietic stem cell transplantation

Guidelines, care pathways, and patient journeys

- 2 guidelines written by ERN TRANSPLANT-CHILD.
- 2 Clinical Audits published, and 2 ongoing

Training and education

- 134 webinars organised since 2018
- 17 short exchange programmes since 2018
- 5 TransplantChild Workshops

Clinical Patient Management System (CPMS)

- 293 CPMS case discussions since 2017

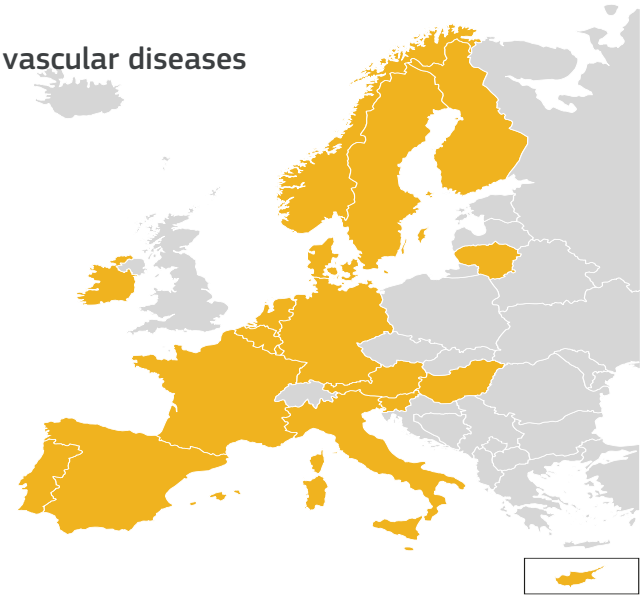
Research and patients' registries

- 6 relevant research proposals involving at least two healthcare providers from two different Member States
- Participation in 8 projects during the last 5 years
- 528 patients enrolled in the PETER registry (https://peter.transplantchild.eu/)
- 11 papers published since 2020

VASCERN

European Reference Network on rare multisystemic vascular diseases

https://vascern.eu/



Members

45 healthcare providers located in 19 countries in the EU and Norway are members of the network, distributed as follows:

- 39 Full Members in 13 Member States, plus Norway
- 4 Associated National Centres in 3 Member States
- 2 National Coordination Hubs in 2 Member States

In addition, the VASCERN European Patient Advocacy Group (ePAG) includes 37 patient representatives and actively collaborates with 75 patient organisations

Disease areas

- Hereditary haemorrhagic telangiectasia
- Heritable thoracic aortic diseases
- Medium sized arteries (vascular Ehlers Danlos Syndrome ...)
- Neurovascular diseases (e.g. Moyamoya disease, CADASIL)
- Paediatric and primary lymphoedema
- Vascular anomalies (malformations, fistulas)

Guidelines, care pathways, and patient journeys

- 12 guidelines co-authored and 30 endorsed by ERN VASCERN
- 10 care pathways
- 14 consensus statements
- 11 sets of Dos and Don'ts factsheets

Training and education

- 19 webinars organised since 2020
- 157 pills of knowledge produced since 2018
- 2 Erasmus+ Summer School organised since 2024
- 53 short exchange programmes
- 3 e-learning courses

Clinical Patient Management System (CPMS)

- 274 CPMS case discussions since 2017

Research and patients' registries

- 24 research projects or clinical trials involving at least two healthcare providers from two different Member States
- 3 030 patients registered in the VASCERN registry (https://vascern.eu/group/registry/)

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