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‘Thousands of patients will benefit’

Vytenis Andriukaitis, European Commissioner for Health and Food Safety, says the value of EU collaboration is particularly clear in the case of rare and complex diseases.

What inspired the creation of the European Reference Networks?

We often hear tragic stories of patients with rare or complex, life-threatening diseases, who face challenges in obtaining a correct diagnosis and in accessing appropriate therapies and clinical expertise. Their doctors are unable to help them as they have never seen similar cases, so they are left untreated or have to scour the internet in the hope of finding a centre with the necessary expertise.

How can ERNs improve the lives of Europeans?

With ERNs, patients with rare and complex conditions will be able to benefit from the best treatment and advice available in the EU for their specific condition. Their doctors will have access to a highly specialised pool of colleagues from all over Europe.

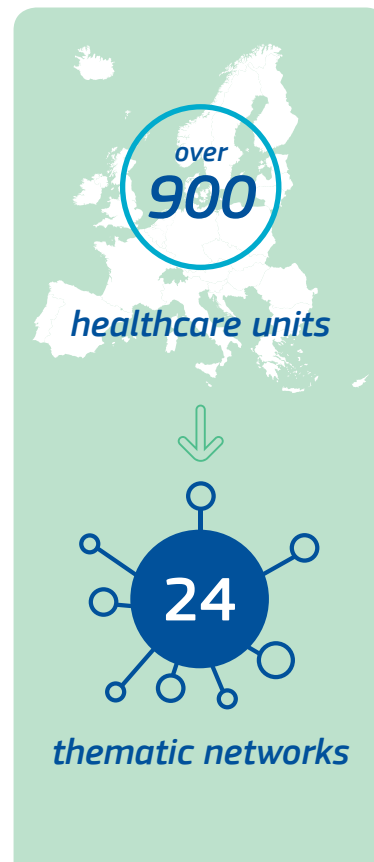
In the first phase, over 900 healthcare units from nearly all EU Member States will work

together in 24 thematic networks. They will cover a wide range of conditions, from bone disorders to blood diseases, from childhood cancer to immunodeficiency. They will facilitate access to diagnosis, treatment and provision of affordable, high-quality and cost-effective healthcare.

What is the added value of EU-level collaboration in this area?

With knowledge and resources on specific rare conditions scattered across individual countries, the EU can provide significant added value by connecting the dots, bringing together expertise and maximising synergies between Member States.

No country alone has the knowledge and capacity to treat all rare and complex conditions, but by cooperating and exchanging life-saving knowledge at European level through ERNs, patients across the EU will have access to the best expertise available.



*'No country alone has
the knowledge and
capacity to treat all rare
and complex conditions'*

Vytenis Andriukaitis

What are the roles of those involved in ERNs?

The driving forces behind the ERNs are health-care providers and national health authorities. They show trust, take ownership and have the most active role in the development and functioning of the networks.

The Commission's role, as defined in the 2011 EU Directive on Patients' Rights in Cross-border Healthcare, is to create the framework for the ERNs. The Commission also provides grants to support network coordinators and provides them with the technical networking facilities.

What additional actions are you taking to tackle rare and complex diseases?

The ERNs are part of a broader strategy to make the national and European health systems more efficient, accessible and resilient. The European Commission supports Member States by pooling knowledge and expertise,

registries, data and funding. We support research and innovation, and fund projects and joint actions. We give incentives to manufacturers to develop orphan medicines and bring them to market.

What is your hope for the future of ERNs?

I hope that ERNs will provide concrete results for tens of thousands of rare disease patients so that they are no longer looking for answers in the dark, and can benefit from the best expertise available in Europe so they may live longer and healthier lives.





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Background

Rare and complex diseases cause chronic health problems and are often life-threatening.

Between **5 000 and 8 000 rare diseases** affect the daily lives of around **30 million people** in the EU. For example, in the field of oncology alone, there are almost **300 different types of rare cancers** and each year more than **half a million people** in Europe are diagnosed with one of them.

Many of those affected by a rare or complex condition do not have access to diagnosis and high-quality treatment. Expertise and specialist knowledge may be scarce because patient numbers are low.

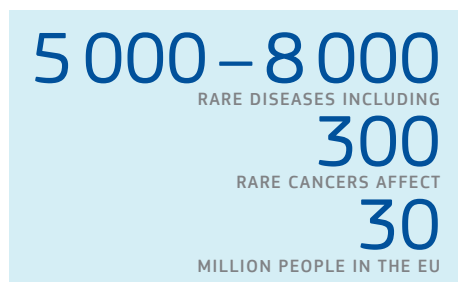
The EU and national governments are committed to improving the recognition and treatment of these rare and complex conditions by strengthening European-level cooperation and coordination and supporting national plans for rare diseases.

The 2011 Directive on Patients' Rights in Cross-border Healthcare not only enables patients to be reimbursed for treatment in another EU Member State but also makes it easier for patients to access information on healthcare and thus increase their treatment options. The Directive became law in

EU Member States in 2013 and emphasises the value of eHealth and the importance of interoperability in national health IT systems in facilitating information sharing.

It is against this backdrop that, with the support of the EU Health Programme, the first 24 European Reference Networks started their activities in 2017.

Many of those affected by a rare or complex condition do not have access to diagnosis and high-quality treatment. Expertise and specialist knowledge may be scarce because patient numbers are low.



What are ERNs?

European Reference Networks (ERNs) are virtual networks involving healthcare providers across Europe. They aim to tackle complex or rare diseases and conditions that require highly specialised treatment and a concentration of knowledge and resources.

To review a patient's diagnosis and treatment, ERN coordinators convene 'virtual' advisory boards of medical specialists across different disciplines, using a dedicated IT platform and telemedicine tools.

No country alone has the knowledge and capacity to treat all rare and complex diseases. ERNs offer the potential to give patients and doctors across the EU access to the best expertise and timely exchange of life-saving knowledge, without having to travel to another country.

Following the first call for proposals in July 2016, the first ERNs were approved in December 2016 and launched in March 2017 in Vilnius where their kick off meetings took place. At their inception, the networks comprised **more than 900 highly specialised healthcare units located in 313 hospitals in**

25 Member States (plus Norway). 24 ERNs are working on a range of thematic issues, including bone disorders, childhood cancer and immunodeficiency. Over the next 5 years, ERNs are expected to reinforce their capacities to benefit thousands of EU patients suffering from a rare or complex condition. Calls for healthcare providers wanting to join existing ERNs will be launched yearly.

The ERN initiative receives support from several EU funding programmes, including the Health Programme, the Connecting Europe Facility and Horizon 2020.

EU Member States lead the ERN process: they are responsible for the recognition of centres at national level; they endorse the applications; and a Board of Member States is responsible for developing the EU ERN strategy and approving the networks.

ERN on endocrine conditions (Endo-ERN)



Rare endocrine conditions include too much, too little or inappropriate hormonal activity, hormone resistance, tumour growth in endocrine organs, or diseases with consequences for the endocrine system. The epidemiological distribution is highly variable from ultra-rare, rare, to low-prevalence conditions. Patients with a low-prevalence disorder may require highly specialised care from a multidisciplinary team led by an endocrinologist.

Endo-ERN has established eight main thematic groups covering the full spectrum of congenital and acquired conditions. These are: adrenal disorders; disorders of calcium and phosphate homeostasis; disorders of sex development and maturation; genetic disorders of glucose and insulin homeostasis; genetic endocrine tumour syndromes; disorders of growth and genetic obesity syndromes; pituitary disorders; and thyroid disorders.

The ERN builds on the work of several existing European networks, including those established through the European Society



Endo-ERN aims to deliver improved diagnostic trajectories, treatment, quality of care and measurable outcome for patients.

of Endocrinology (ESE) and European Society for Paediatric Endocrinology (ESPE), and those developed through COST Actions.

Endo-ERN aims to deliver improved diagnostic trajectories, treatment, quality of care and measurable outcome for patients with rare endocrine conditions by facilitating multidisciplinary and cross-border collaboration and education and by listening to the patient. ■

NETWORK COORDINATOR

Professor Alberto M. Pereira
*Leiden University Medical Center,
The Netherlands*

ERN on kidney diseases (ERKNet)



Rare and complex kidney diseases comprise a wide range of congenital, hereditary and acquired disorders. It is estimated that at least 2 million Europeans are affected by rare kidney diseases, with glomerulopathies and congenital kidney malformations each accounting for approximately 1 million cases. In addition, inherited tubulopathies, tubulointerstitial diseases and thrombotic microangiopathies represent a number of rare and ultra-rare diseases of high clinical relevance.

State-of-the-art diagnostic tools can provide valuable information about disease prognosis and therapeutic options. However, access to testing is not universal. Due to delayed diagnosis and delayed treatment, many rare kidney diseases progress to renal failure.

This ERN will seek to improve standards of diagnosis and treatment across Europe. The network will establish a consensus on rational diagnostic algorithms for patients presenting with signs and symptoms of renal disease, including standard criteria for genetic testing

Online consultation services will improve management of new and complex cases.

in cases of suspected hereditary kidney disease. Working groups will then define clinical pathways for therapeutic management after thorough analysis of available treatments.

Online consultation services will improve management of new and complex cases. Access to a virtual consultation board will be complemented by administrative measures to facilitate patient travel to specialised centres where necessary, in line with the EU Cross-border Healthcare Directive and Social

Security Regulation. A series of webinars will be developed for teaching and training health professionals.

NETWORK COORDINATOR

Professor Franz Schaefer
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Germany*

Adding value for patients and professionals

Patients with rare and complex diseases can spend years without a clear diagnosis. It can be a frustrating and dispiriting experience for patients and their families. Many people living with these conditions are children whose development is severely affected as they move through the health system, sometimes seeing several specialists, in search of a diagnosis.

ERNs improve public and professional awareness of rare diseases and complicated presentations of illness, increasing the likelihood of early and accurate diagnosis and effective treatment where available.

The networks are a platform for the development of guidelines, training and knowledge-sharing. ERNs can facilitate large clinical studies to improve understanding of diseases and develop new drugs by gathering a large pool of patient data.

For specialist health professionals, the ERNs are an opportunity for networking with like-minded experts from across Europe — ending the professional isolation that many experts in rare diseases face.

Innovation in healthcare delivery is the cornerstone of the ERN system with the development of new care models, eHealth solutions and tools, and innovative medical solutions and devices, changing the way in which treatment itself is delivered. ERNs are incubators for the development of digital services for the provision of virtual healthcare.

ERNs will help to boost economies of scale and ensure a more efficient use of resources, with a positive impact on the sustainability of national healthcare systems. The networks are a visible demonstration of what solidarity can achieve in Europe.

The networks are a platform for the development of guidelines, training and knowledge-sharing.



ERN on bone disorders (ERN BOND)



Rare bone diseases encompass disorders of bone formation, modelling, remodelling and removal, and defects of the regulatory pathways of these processes. They result in short stature, bone deformity, teeth anomalies, pain, fractures and disability, and can adversely influence neuromuscular function and haemopoiesis.

ERN BOND brings together all rare bone diseases — congenital, chronic and of genetic origin — that affect cartilage, bones and dentin. The network is focusing initially on osteogenesis imperfecta (OI), X-linked hypophosphataemic rickets (XLH) and achondroplasia (ACH) as exemplars, based on disease prevalence, diagnostic and management difficulty and novel emergent therapy, before moving on to rarer diseases when systematic approaches are established.

Working with patients, BOND will develop patient-reported outcome and experience measures. The network will develop guidelines, leading to the development and dissemination of best practice. As new therapeutics are



*Working with patients,
BOND will develop
patient-reported outcome
and experience measures.*

developed, the network will work to ensure rapid access to studies for affected patients.

BOND will enable skill development through eHealth and telemedicine platforms, alongside working visits, training courses and dissemination activities. The network aims to reduce time to diagnosis with fewer inappropriate tests, more accurate diagnosis and new viable treatments to be available within 2 to 3 years.

NETWORK COORDINATOR

Dr Luca Sangiorgi
*Rizzoli Orthopaedic Institute,
Bologna, Italy*

How ERNs are approved

EU Member States play the lead role in the designation and development of European Reference Networks. To achieve ERN status, network members applied to a Call from the European Commission. This application was assessed by an Independent Assessment Body (IAB) which completed reports on each applicant. The Board of Member States (BoMS) then decided whether or not to approve an ERN application.

The BoMS comprises nominees from all EU Member States plus Norway and plays an active role in developing the ERN strategy.

The BoMS continues to monitor ERN members, assess applicants wishing to join existing networks, and approve any future networks.

Countries which do not have representation in an approved ERN may participate through healthcare providers that are designated by their Member State as 'associated' and/or 'collaborative' national centres.

Key criteria

Patient-centred and clinically led

10 members in at least
8 countries

Strong independent assessment

Fulfilment of Network
and Member criteria

Endorsement and approval
by national authorities.

'This brings practical benefits in terms of patient care and in managing the networks.'

Professor Katarzyna Kotulska-Jóźwiak, a consultant neurologist and one of Poland's representatives on the BoMS says experts and patients were consulted when determining the composition of ERNs. 'We wanted to have networks for individual disease areas in order to meet the expectations of stakeholders,' she says. 'This brings practical benefits in terms of patient care and in managing the networks.'

ERN on craniofacial anomalies and ENT disorders (ERN CRANIO)



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Congenital craniofacial anomalies include children born with underdeveloped or maldeveloped parts of their brain, skull and/or face that result in significant functional problems and psychosocial challenges. Patients require follow-up and treatment from birth into adulthood. Clinical and public knowledge of many of these presentations is low, and diagnosis can be extremely challenging.

This ERN addresses several gaps in care by significantly improving familiarity of primary caregivers with craniofacial anomalies. The network is developing instructional courses on numerous conditions to be made available through an open access website.

Members are working together to improve education, training and research in close collaboration with patient organisations. Where no patient organisation exists, focus groups of patients are consulted. ERN CRANIO is evaluating the type and timing of surgical



The network is developing instructional courses on numerous conditions to be made available through an open access website.

treatment at participating centres to shed light on their impact and benchmark best practices in Europe.

By collecting data on long-term outcomes of the various conditions, the network will aid in counselling patients and parents and can direct the focus of treatment to areas that have received too little attention. The network will support the detection of new causative genes by increasing the numbers of participants in research studies.

NETWORK COORDINATOR

Professor Irene Mathijssen
*Erasmus MC: University Medical Center
Rotterdam, The Netherlands*

ERN on epilepsies (EpiCARE)

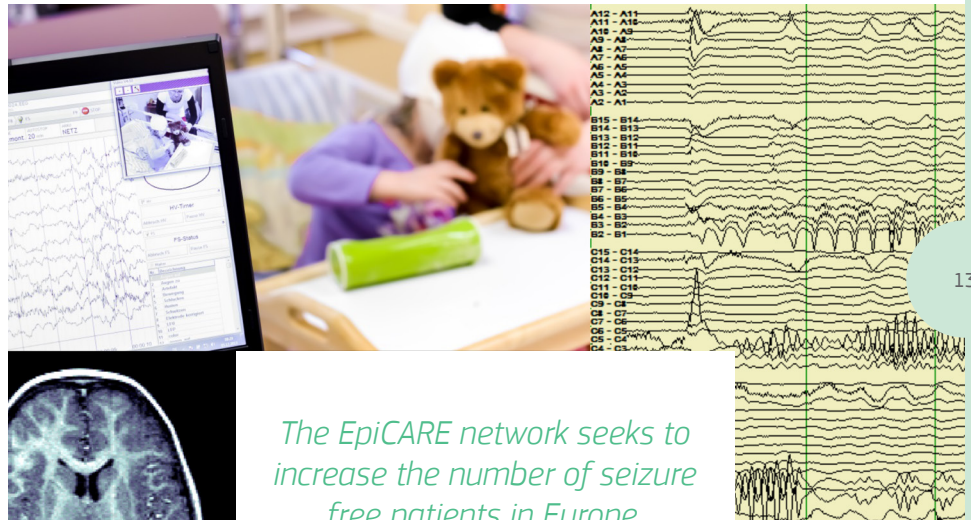


Epilepsy affects at least 6 million people in Europe. Traditional antiepileptic therapies help between **60% and 70%** of those affected to remain **seizure free**. For patients suffering from refractory epilepsy, the clinical outlook is poor.

Traditionally, epilepsy has been treated as a single disease, but these conditions are increasingly viewed as a group of rare and complex diseases. ORPHANET — the portal for rare diseases and orphan drugs — lists 137 disorders with epilepsy as the predominant symptom, however many patients remain undiagnosed and without access to treatment.

The network aims: to deliver full access and utilisation of pre-surgical evaluation and epilepsy surgery; to increase diagnosis of rare causes of the epilepsies; to enhance identification of patients with treatable rare causes of the epilepsies; to increase access to specialised care for rare causes; and to foster research on innovative causal treatments in rare and complex epilepsies.

EpiCARE builds on the work of the pilot ERN E-epilepsy which worked to increase awareness and accessibility of epilepsy surgery, for carefully selected individuals, that effectively



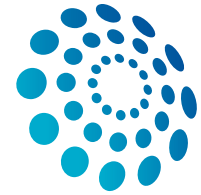
The EpiCARE network seeks to increase the number of seizure free patients in Europe.

used e-tools and multidisciplinary team discussion. The EpiCARE network, which includes active participants from patient organisations, seeks to increase the number of seizure free patients in Europe.

NETWORK COORDINATOR

Alexis Arzimanoglou
Centre Hospitalier Universitaire de
Lyon, CHU Lyon, France

ERN on adult cancers (solid tumours) (ERN EURACAN)



More than **300 rare cancers** have been identified. ERN EURACAN covers all rare adult solid tumour cancers, grouping them into 10 domains corresponding to the RARECARE classification and ICD10. The management of rare cancers poses significant diagnostic challenges, sometimes with major consequences for patients' quality of life and outcome. Inappropriate management of these patients may also result in an increased risk of relapse, and risk of death.

The network aims to reach all EU countries within 5 years and develop a referral system to ensure at least 75% of patients are treated in a EURACAN centre.



ERN EURACAN is sharing best practice tools and establishing reference centres for rare cancers. It is also establishing regularly updated diagnostic and therapeutic clinical practice guidelines. The network aims to reach all EU countries within 5 years and develop a referral system to ensure at least 75% of patients are treated in a EURACAN centre. It seeks to improve patient survival, produce communication tools in all languages for patients and physicians, and develop multinational databases and tumour banks.

The ERN builds on pre-existing clinical and research networks that have successfully conducted clinical trials through the European Organisation for Research and Treatment of Cancer (EORTC), and established guidelines through EORTC and the European Society for Medical Oncology (ESMO). It also benefits from the work of networks formed by the European Neuroendocrine Tumour Society (ENETS) and Connective Tissues Cancer Network (Conticanet), as well as several EU research projects. ■

NETWORK COORDINATOR

Professor Jean-Yves Blay
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Member States in the driving seat

Paul Boom represents the Netherlands on the Board of Member States (BoMS). **The BoMS is the key player in determining the future of ERNs and in approving the networks.** 'The legislation makes clear that Member States are in the driving seat,' he says. 'It is the national authorities that determine whether ERN applicants fulfilled the criteria in terms of quality, patient involvement and governance.'

At national level, Member States also work to ensure ERNs are well connected to national health services, according to Mr Boom. 'ERNs must not become freestanding islands of excellence operating in isolation,' he says. 'They have good links to hospital and primary care services and benefit communities in their locality as well as supporting patients around Europe.'

Looking more broadly at the impact of ERNs, Mr Boom says the networks mark an exciting new chapter in European cooperation in the field of healthcare. They are a clear example of how Member States can work together to add value for citizens. 'In my own view, ERNs could serve as a platform for the development of eHealth tools and could even see greater cooperation on more common chronic illnesses,' he said. 'We now have a platform on which to build; an opportunity for Member States to come together to speak about shared challenges in healthcare and to think beyond borders.'



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ERN on haematological diseases (EuroBloodNet)



Haematological diseases involve abnormalities of blood and bone marrow cells, lymphoid organs and coagulation factors, and almost all of them are rare. They can be subdivided into six categories: rare red blood cell defects; bone marrow failure; rare coagulation disorders; haemochromatosis and other rare genetic disorders of iron synthesis; myeloid malignancies; and lymphoid malignancies.

Diagnosis of rare haematological diseases (RHDs) requires considerable clinical expertise and access to a broad range of laboratory services and imaging technologies. These tests allow precise disease classification according to WHO criteria using international scoring systems and, where possible, biomarkers.

Given these requirements and the fact that some RHDs are very rare, diagnosis is frequently overlooked or delayed, especially in elderly patients. Treatment is also often difficult due to the specialised infrastructures and teams required and the difficulty accessing specific treatments such as allogenic stem cell transplantation or coagulation factors.

Preventive programmes are in place in some countries for certain conditions, but there is an urgent need for harmonisation in the field of screening.

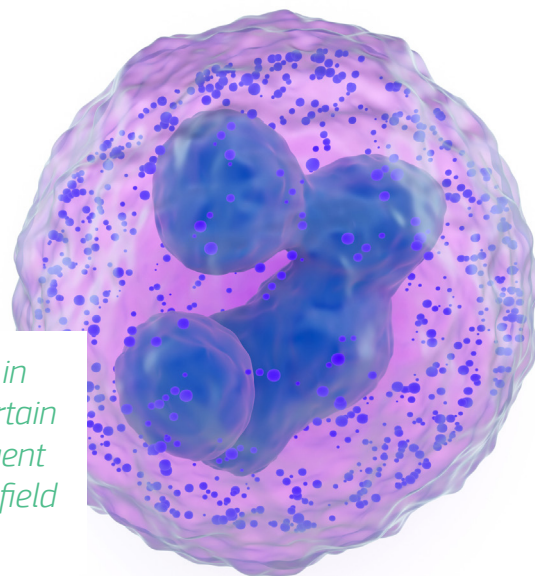
Preventive programmes are in place in some countries for certain conditions, but there is an urgent need for harmonisation in the field of screening.

EuroBloodNet, with the experience gained thanks to the EU-funded European Network for Rare and Congenital Anaemias (ENERCA) and the European Haematology Association (EHA), will seek: to improve access to healthcare for RHD patients; to promote guidelines and best practice; to improve training and knowledge-sharing; to offer clinical advice where

national expertise is scarce; and to increase the number of clinical trials in the field.

NETWORK COORDINATOR

Professor Pierre Fenaux
Assistance Publique-Hôpitaux de Paris,
Hôpital Saint-Louis, France





ERN on urogenital diseases and conditions (ERN eUROGEN)

Rare and complex urogenital conditions can require surgical correction, often during the neonatal period or in childhood. Urinary and faecal incontinence are a heavy burden on paediatric, adolescent and adult patients. Individuals affected require life-long care provided by multidisciplinary teams of experts who plan and perform surgery, and provide post-operative physiotherapy and psychological support.

eUROGEN will provide independently-evaluated best practice guidelines and improve the sharing of outcomes. It will, for the first time, offer the capacity for tracking long-term outcomes for patients over a 15 to 20-year period.

The network will collect data and materials where they are lacking, develop new guidelines, build evidence of best practice, identify practice variation, develop education programmes and training, set the research agenda in collaboration with patient representatives, and share knowledge through participation in virtual multidisciplinary teams. By 2020, at least 50 new specialists for rare



and complex urogenital diseases will have benefited from specific training and fellowship programmes developed by eUROGEN.

Ultimately, the network seeks to advance innovation in medicine and improve diagnostics and treatment for patients.

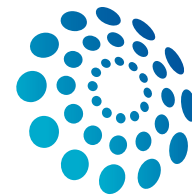
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NETWORK COORDINATOR

Wout Feitz

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ERN on neuromuscular diseases (ERN EURO-NMD)



Neuromuscular diseases (NMDs) occur from early childhood to late adulthood and are characterised by muscle weakness and wasting, but may be associated with other symptoms, including fatigue, pain, numbness, blindness, swallowing difficulties, breathing difficulties and heart disease. Most NMDs are progressive and debilitating, with reduced lifespan and quality of life.

There are significant gaps and disparities in access to diagnostics and treatment across Europe. Major challenges in improving outcomes include the delay in referral from primary care to a specialist centre, and managing the transition from paediatric to adult services.

ERN EURO-NMD unites Europe's leading experts to provide patients with access to specialist care through virtual and in-person consultations. The network aims to reduce time to diagnosis by 40% in its first 5 years, to improve diagnostic yield by 15% and to increase access to appropriate care pathways.



The network aims to reduce time to diagnosis by 40% in its first 5 years, to improve diagnostic yield by 15% and to increase access to appropriate care pathways.

In addition, ERN EURO-NMD will develop new guidelines and provide healthcare professionals and patients with disease-specific best-practice information. The knowledge generated and curated by the network will be widely available through eHealth tools. Building on a strong legacy of cooperation, the network will also foster collaborations with the potential to drive research and therapy development to address unmet patient needs.

NETWORK COORDINATOR

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Europe: a global centre of excellence

European Reference Networks came into operation in March 2017. While these networks are still new and their primary purpose is to improve the lives of people in Europe who are living with rare and complex diseases, they will have a global impact.

ERNs will tap into global best practice where it exists, and create it where it does not. **The networks will help to make Europe a hub of activity in rare and complex conditions.**

For example, ERNs will be well-placed to implement best practice guidelines. For conditions where no diagnostic or treatment guidelines exist, the networks may have the capacity to develop guidance and best practices.

By connecting experts and patient populations, ERNs also have the potential to facilitate clinical studies and test therapeutic interventions. This will put them at the forefront of innovation in numerous rare disease fields.

The ERN model could become an example to others. The state-of-the-art eHealth tools developed to aid cross-border collaboration in



Europe may also have the potential to foster international collaborations, while improving healthcare access.

ERNs will tap into global best practice where it exists, and create it where it does not.

ERN on eye diseases (ERN EYE)



Rare Eye Diseases (RED) are the leading cause of visual impairment and blindness for children and young adults in Europe. More than 900 REDs are listed in the portal for rare diseases and orphan drugs (ORPHANET). These include more prevalent diseases such as retinitis pigmentosa which has an estimated prevalence of 1 in 5 000, as well as some very rare entities described only once or twice in medical literature.

ERN EYE addresses these conditions in four thematic groups: rare diseases of the retina, neuro-ophthalmology rare diseases, paediatric ophthalmology rare diseases, and rare anterior segment conditions.

In addition, six transversal working groups are addressing issues common to the four main themes. Additional working groups focus on specific areas, including genetic testing, registries, research, education, communication and patients.

The network's main aim is the development of a virtual clinic — known as EyeClin — to guarantee the best coverage of REDs and facilitate cross-border dissemination of expertise.

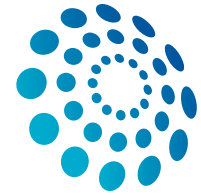
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NETWORK COORDINATOR

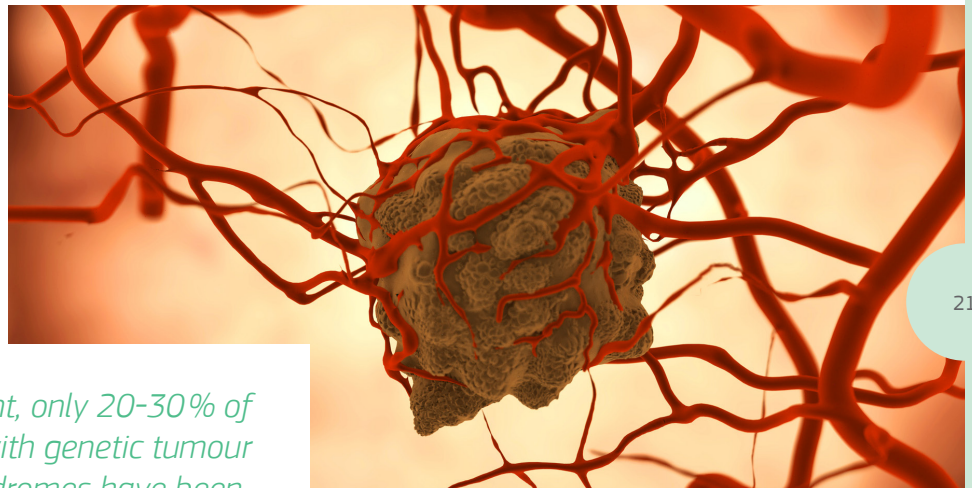
Professor H el ene Dollfus
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France

ERN on genetic tumour risk syndromes (ERN GENTURIS)



Genetic tumour risk syndromes are disorders in which inherited genetic mutations strongly predispose individuals to the development of tumours. The lifetime risk of cancer can be as high as 100%. While there is considerable diversity in the organ systems that may be affected, individuals affected by these conditions share similar challenges: delay in diagnosis, lack of prevention for patients and healthy relatives, and therapeutic mismanagement. At present, only 20-30% of people with genetic tumour risk syndromes have been diagnosed.

ERN GENTURIS is working to improve identification of these syndromes, minimise variation in clinical outcomes, design and implement guidelines, develop registries and biobanks, support research, and empower patients. The network will educate the public and healthcare professionals, and foster the sharing of best practice across Europe. Access to multidisciplinary care will be improved, with new models and standards for sharing and discussing complex cases. The network is enhancing the quality and interpretation of genetic testing,



At present, only 20-30% of people with genetic tumour risk syndromes have been diagnosed.

and increasing patient participation in clinical research programmes.

ERN GENTURIS will cooperate with other ERNs to improve the care of patients with genetic tumour risk syndromes who develop conditions that fall within the expertise of another network.

NETWORK COORDINATOR

Prof. Nicoline Hoogerbrugge
Radboud University Medical
Center Nijmegen, The Netherlands

Collaboration in action

Information technology (IT) and eHealth tools can play a valuable role in facilitating collaboration. ERNs are connected through a dedicated IT platform through which a network coordinator can convene 'virtual' advisory boards of medical specialists using telemedicine tools to review a patient's condition for diagnosis or treatment. This allows health professionals, who would previously have handled rare and complex cases in isolation, to consult peers and seek a second opinion from a colleague. A central feature of these tools is interoperability.

Thanks to technology, geography need not be a barrier to working in dispersed teams. In some cases, phone or video calls will suffice. On other occasions, networks can use dedicated systems to share tissue samples or high-resolution images of complex conditions. These technologies can also be used as a repository of cases, helping to build a large bank of cases for further study.

For example, once pathology or radiology data are securely shared, members of the network can log in, view the images and comment

in a closed environment. The treating physician remains responsible for their patient, but can tap into the ERN as a valuable and supportive resource.

Thanks to technology, geography need not be a barrier to working in dispersed teams.

Affiliated Partners

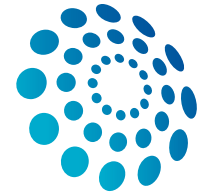
ERNs aim to deliver genuine added value to all EU Member States. The relevant legislation enables **countries without representation in an approved ERN** to participate through healthcare providers that are designated by their Member State as 'associated' and/or 'collaborative' national centres.

Member States may also wish to designate a national coordination hub to liaise with all ERNs. The ERN Board of Member States sets up the common framework for the designation and integration of those types of centres into the ERNs. Nevertheless, it is essential that the designation of Affiliated

Partners by Member States be undertaken through open, transparent and robust procedures.

The first affiliated partners should be nominated by some of the Member States by the end of 2017.

ERN on diseases of the heart (ERN GUARD-HEART)

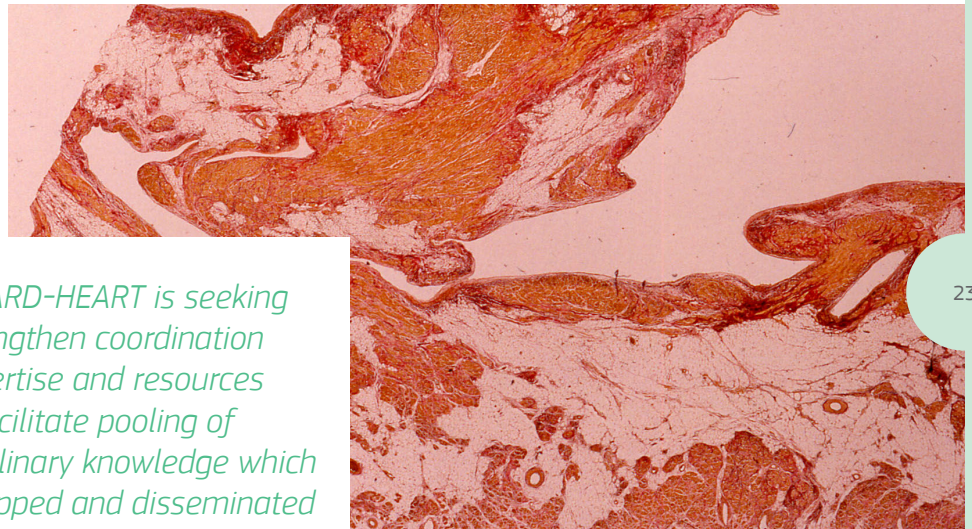


Rare cardiac diseases can present throughout a person's life and are mostly genetic. These conditions are characterised by a wide range of symptoms and signs that vary not only from disease to disease but also from patient to patient. All these cardiac diseases carry a unique susceptibility to sudden death at a young age, usually occurring in otherwise healthy people.

The GUARD-HEART network has identified the following thematic areas: familial electrical diseases, familial cardiomyopathies, congenital heart defects and other rare cardiac diseases. These themes are based

on the clinical guidelines of the European Society of Cardiology (ESC), the International Classification of Diseases (ICD10) and ORPHANET.

ERN GUARD-HEART is seeking to strengthen coordination of expertise and resources to facilitate pooling of multidisciplinary knowledge which will be mapped and disseminated to the lay public.



ERN GUARD-HEART is seeking to strengthen coordination of expertise and resources to facilitate pooling of multidisciplinary knowledge which will be mapped and disseminated to the lay public.

Healthcare services will be provided through a shared eHealth platform. This will ensure wider access to expertise for patients and healthcare professionals around Europe. By fostering closer cooperation between experts, new scientific knowledge will be acquired and shared to support the development of new diagnostic and therapeutic procedures, and to identify new rare cardiac diseases. ■

NETWORK COORDINATOR

Professor Arthur Wilde
*Academic Medical Center,
Amsterdam, The Netherlands*

ERN on inherited and congenital abnormalities (ERNICA)



ERNICA addresses congenital malformations and diseases that appear early in life and require multidisciplinary care and long-term follow-up, and examines the transition to adulthood.

The network is organised around two main work streams in line with ORPHANET classifications and ICD10. One work stream deals with malformations of the digestive system and the other deals with malformations of the diaphragm and abdominal wall. In the latter work stream, there are working groups covering malformations of the oesophagus and a group working on gastroenterological and intestinal diseases. This group also includes a sub-group specialising in intestinal failure. Each working group has its own disease-specific task forces.

For some of these rare diseases, mortality rates can be as high as 50%. ERNICA aims to improve the quality of care that patients receive and to reduce the long-term impact of these rare diseases in infants. The network will facilitate research collaborations with the power to develop evidence-based clinical



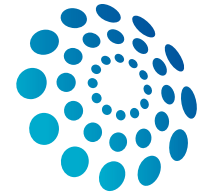
For some of these rare diseases, mortality rates can be as high as 50%.

guidelines. Access to new surgical techniques and treatments will also be improved.

ERNICA is a meeting place for national patients' associations and caregivers, including nurses and other professions committed to improving patient outcomes.

NETWORK COORDINATOR

Professor Rene Wijnen
Erasmus Medical Center Rotterdam,
The Netherlands



ERN on congenital malformations and rare intellectual disability (ERN ITHACA)

This ERN brings together experts in rare congenital malformations and rare intellectual disability disorders. Congenital malformations affect one in 40 babies. For more common malformations, such as cleft lip, there are well-established care networks. For rarer conditions, expertise is scattered across the EU. Many malformations occur together as part of 'syndromes' associated with abnormal growth, development or social adaptation. Over **8 000 syndromes** have been described, and most occur at a frequency of **less than 1 in 2 000**.

Chromosome disorders are one of the commonest causes of malformations and intellectual disability. New tests, such as exome and genome sequencing, have improved the prospects of diagnosis but are not routinely available in more than 50% of highly specialised centres.

Expanding access to this technology is a key goal of ERN ITHACA. The network is also developing telehealth initiatives with virtual



multidisciplinary teams across EU centres, and will use virtual online clinics to improve access to diagnostics without requiring patients to travel.

ERN ITHACA will network parents and patients to develop best practice and initiate guideline development where required. It will establish criteria for patient registry data, advance training for health professionals and facilitate research. The network will work with existing networks in the field and with ERNs

with whom there are complementary interests, while keeping patients at the centre of its activities.

NETWORK COORDINATOR

Alain Verloes

*Assistance publique - Hôpitaux de Paris,
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Leading a European Reference Network

Professor Pierre Fenaux, Professor of Haematology at Hôpital Saint-Louis, Paris, France, leads the EuroBloodNet ERN. For Prof. Fenaux, the 66-member EuroBloodNet network offers significant benefits to patients and professionals. 'The purpose of the ERN is to be patient-centred; to improve access to healthcare in rare haematological disorders,' he says. 'We are bringing cutting-edge diagnostics and treatments to centres around Europe where the required expertise may not exist.'

He says the endorsements of EU Member States and the European Commission add weight to the network and improve its capacity for disseminating guidelines. 'We also see great opportunities for teaching about rare haematological diseases through continuing medical education (CME) systems,' Prof. Fenaux adds.

For clinicians, the everyday benefit of the network comes when dealing with rare or complex cases: 'Physicians can consult the expertise of colleagues in other countries — this ends the isolation that health professionals sometimes feel if they do not have access to peers who can give a second opinion.'

There are other potential benefits too. Prof. Fenaux says that connecting hospitals around Europe will create a critical mass of patients with rare diseases, paving the way for clinical research which would previously not have been feasible.

These linkages may also serve as an advocacy platform by fostering the development of patient associations for people with rare diseases, and offering expert input on innovative treatments. 'If a local physician asks their hospital for access to an innovative treatment, our network could offer expert opinion on the science behind a new intervention,' says Prof. Fenaux. 'Physicians and patients in this field now know that they are not alone.'



'Physicians can consult the expertise of colleagues in other countries — this ends the isolation that health professionals sometimes feel if they do not have access to peers who can give a second opinion.'



ERN on respiratory diseases (ERN LUNG)

Complex lung diseases require multidisciplinary care along with psycho-social support. This complexity can be due to the underlying genetic mechanism of the disease, the secondary changes and damage done to other organ systems. Early diagnosis and access to specialist care can improve outcomes for many of these conditions.

ERN-LUNG addresses a number of rare and complex pulmonary conditions, including idiopathic pulmonary fibrosis, cystic fibrosis, non-cystic fibrosis bronchiectasis, pulmonary hypertension, PCD, AATD, mesothelioma, chronic lung allograft dysfunction, and ORLD.

The network seeks to improve expertise across Europe to advance standards of care, quality of life and prognosis across the spectrum of rare pulmonary diseases. Members are: developing and disseminating care guidelines; promoting common treatment approaches; enhancing cross-border access to diagnosis and treatment; initiating and supporting registries; and assembling sufficiently large



The network seeks to improve expertise across Europe to advance standards of care, quality of life and prognosis across the spectrum of rare pulmonary diseases.

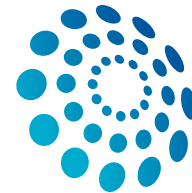
cohorts for clinical studies, drug development and natural history studies.

ERN-LUNG provides patients with access to the interdisciplinary teams, providing online second opinions on complex cases without requiring patients to travel. This will see the expansion of an online expert advice system established through the EU-funded pilot project, ECORN-CF.

NETWORK COORDINATOR

Professor Thomas O.F. Wagner
Universitätsklinikum Frankfurt,
Germany

ERN on paediatric cancer (haemato-oncology) (ERN PaedCan)



Paediatric cancer is rare and comes in multiple subtypes. With 20 000 children newly diagnosed with cancer across Europe and 6 000 paediatric cancer patients dying each year, it remains the leading cause of death from disease for children older than 1 year of age.

Average survival rates have improved in recent decades; for some conditions the progress has been dramatic, while for others the outcomes remain very poor. Significant inequalities in survival rates are also a challenge in Europe, with worse outcomes in Eastern Europe.

ERN PaedCan is working to improve access to high-quality healthcare for children with cancer whose conditions require specialist expertise and tools not widely available due to low case volumes and a lack of resources. It builds on previous EU-funded projects ENCCA, PanCare and ExPO-r-Net. ERN PaedCan is building a roadmap of specialist centres to



A paediatric oncology tumour board network will be implemented using IT tools to share expertise and advice.

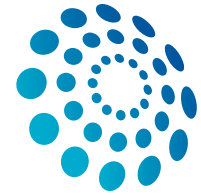
help improve their visibility to healthcare providers and patients. A paediatric oncology tumour board network will be implemented using IT tools to share expertise and advice.

The network aims to increase childhood cancer survival and quality of life by fostering cooperation, research and training, with the ultimate goal of reducing current inequalities in childhood cancer survival and healthcare capabilities in EU Member States.

NETWORK COORDINATOR

Professor Ruth Ladenstein
*St. Anna Kinderspital & St. Anna
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ERN on hepatological diseases (ERN RARE-LIVER)



Rare liver diseases can cause progressive liver injury leading to fibrosis and cirrhosis. The complications of cirrhosis can lead to death and, in many cases, the only effective treatment is liver transplantation. Fatigue, pruritus in cholestatic conditions, and pain and abdominal swelling in cystic conditions significantly affect quality of life.

In paediatric patients, delay in diagnosis, and failure to thrive and attain developmental milestones are additional key factors, along with the challenge of transition in care through adolescence.

ERN RARE-LIVER addresses three disease themes: autoimmune liver disease, metabolic biliary atresia and related liver disease, and structural liver disease. The network will, for the first time in liver disease, fully integrate adult and paediatric care with a focus on the needs of transitional populations and the implications for families with a genetic diagnosis.

The development of up-to-date guidelines is a priority. Care guidelines will be



The network will, for the first time in liver disease, fully integrate adult and paediatric care.

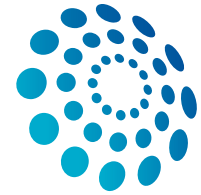
implemented in collaboration with the European Association for the Study of the Liver (EASL) and the European Society for Paediatric Gastroenterology, Hepatology and Nutrition (ESPGHAN). This will be supported by the standardisation of key diagnostic and prognostic tests.

Clinician awareness of rare liver disorders and equitable access to rapidly evolving treatment options are major challenges to be addressed.

NETWORK COORDINATOR

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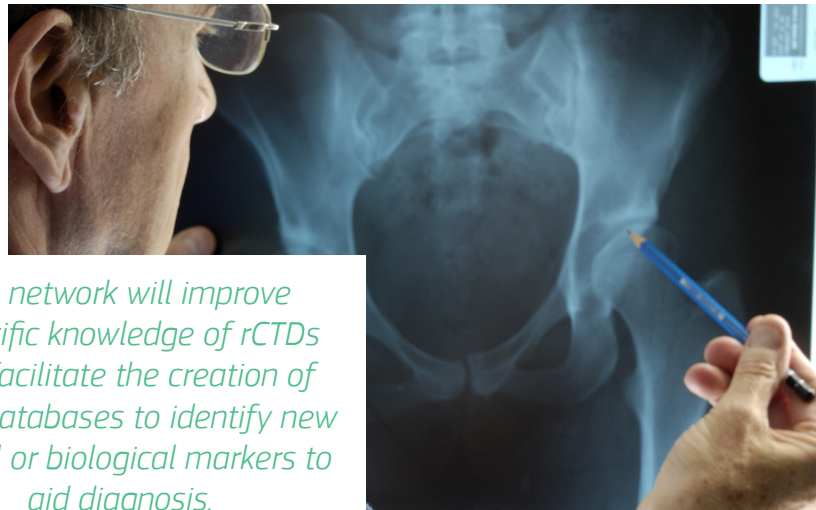
ERN on connective tissue and musculoskeletal diseases (ERN ReCONNET)



Rare connective tissue and musculoskeletal diseases (rCTDs) comprise a large number of diseases and syndromes, with a tremendous impact on patient well-being. These include hereditary conditions, and systemic autoimmune diseases such as systemic sclerosis, mixed connective tissue diseases, inflammatory idiopathic myopathies, undifferentiated connective tissue diseases, and anti-phospholipid syndrome. Delayed diagnosis, particularly for rare or complex presentations, is a common problem.

This network groups rCTDs into three main thematic groups: rare autoimmune, complex autoimmune, and rare hereditary connective tissue and musculoskeletal diseases.

ReCONNET aims to improve early diagnosis, patient management, care delivery and virtual discussion of clinical cases within the network and with affiliated centres. The use of information technologies (IT) will facilitate interaction between centres. The network will improve



The network will improve scientific knowledge of rCTDs and facilitate the creation of large databases to identify new clinical or biological markers to aid diagnosis.

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Educational programmes for patients and families will be developed and disseminated, and new guidelines and quality measures will be implemented. Improved therapeutic protocols and greater patient involvement are also priorities.

NETWORK COORDINATOR

Professor Marta Mosca
*Azienda Ospedaliero Universitaria Pisana,
Italy*

National policies on rare diseases

EU Member States are responsible for national health policy and health service delivery. In 2009, the European Council of Health Ministers¹ recommended that member countries establish and implement plans or strategies by the end of 2013 to support rare diseases patients. According to the recommendation, the plans would be designed to:

- + Guide and structure actions in rare diseases within national health and social systems
- + Integrate initiatives at local, regional and national levels into plans or strategies to ensure a comprehensive approach
- + Define priority actions with objectives and follow-up mechanisms

National plans/strategies implementation has been supported by projects funded from the EU Health Programmes. In 2009, a focus on rare diseases was relatively new

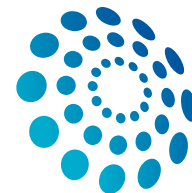


and innovative in most Member States and only four had national plans in place. Currently 23 Member States have adopted their plans/strategies.

http://ec.europa.eu/health/rare_diseases/national_plans/detailed_en

(¹) <http://eur-lex.europa.eu/LexUriServ/LexUriServ.do?uri=OJ:C:2009:151:0007:0010:EN:PDF>

ERN on immunodeficiency, autoinflammatory and autoimmune diseases (ERN RITA)

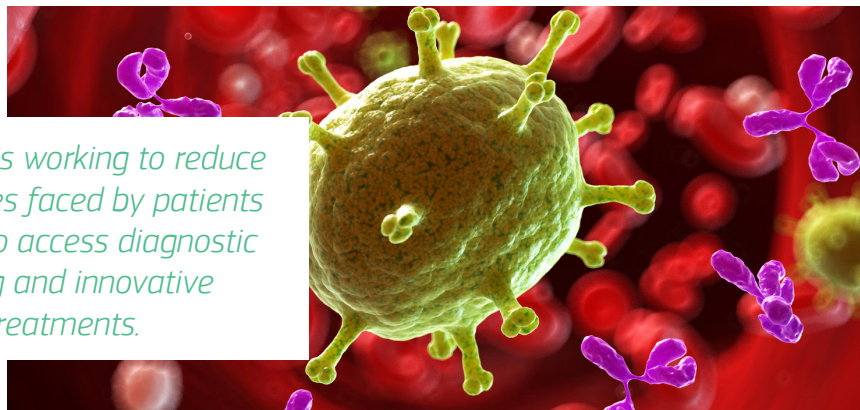


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RITA brings together the leading European centres with expertise in diagnosis and treatment of rare immunological disorders. These constitute potentially life-threatening conditions requiring multidisciplinary care using complex diagnostic evaluation and highly specialised therapies. The network divides these conditions into three sub-themes: primary immunodeficiency (PID), autoimmune disorders and autoinflammatory disorders. In addition, there is a sub-theme of paediatric rheumatology which straddles the autoimmune and auto-inflammatory sub-themes.

This network builds on the work of European scientific societies which have developed patient registries, clinical guidelines, research collaborations, educational activities and links with patient organisations.

ERN RITA is working to reduce inequalities faced by patients seeking to access diagnostic testing and innovative treatments.



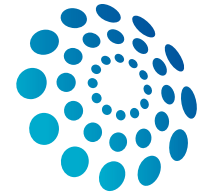
ERN RITA is working to reduce inequalities faced by patients seeking to access diagnostic testing and innovative treatments such as biologic therapies, immunoglobulin replacement, stem cell transplantation and gene therapy.

It aims to link pre-existent registries, develop pan-European clinical guidelines, establish a task force of geneticists for quality control of next generation sequencing technology, agree a common tool for pharmacovigilance in these rare conditions, convene a task force for the correct use and monitoring of biologic treatments in immune-mediated diseases,

bring together and improve stem cell and gene therapies for patients, foster collaborations between patient associations, and bring together paediatric and adult specialists across the three themes.

NETWORK COORDINATOR

Nico Martinus Wulffraat
University Medical Center Utrecht, The Netherlands



ERN on neurological diseases (ERN-RND)

The European Reference Network on Rare Neurological Diseases (ERN-RND) aims to address the unmet needs of more than 500 000 people living with RNDs in Europe. Due to significant phenotype and genotype heterogeneity of RND patients, 60 % of those affected are still undiagnosed.

ERN-RND seeks to address these gaps through virtual multidisciplinary consultation, increasing the number of patients in registries by 20 %, and aims for a 20 % improvement in case outcomes — the percentage of patients with a final diagnosis. Multidisciplinary care pathways will be developed in collaboration with the European Pathway Association and ORPHANET.

The network builds on existing infrastructure by integrating a number of mature RND networks under the ERN-RND umbrella and supplementing functioning registries for conditions such as Huntington's disease and ataxia.



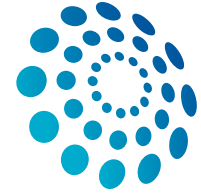
More than 500 000 people living with RNDs in Europe, 60% of those affected are still undiagnosed.

An external quality assessment scheme for the standardisation of key diagnostic tests will be developed in cooperation with the European Molecular Genetics Quality Network, ensuring all patients have access to the same diagnostic opportunities. ERN-RND will support training, research and innovation interventions, and ensure patients' voices are heard. ■

NETWORK COORDINATOR

Dr Holm Graessner
Universitätsklinikum Tübingen,
Germany

ERN on skin disorders (ERN Skin)



Many skin conditions have a severe impact on patients and can be associated with a risk of cancer. Diagnosis of rare and complex skin diseases consists of a full assessment of the skin and mucous membrane, as well as other systems, and skin biopsies. Only experienced dermatologists can differentiate between these complex conditions. The absence of an expert diagnosis is a barrier to treatment. This can be a profound physical and psychological burden for patients.

This network brings together leading experts in the field of rare child and adult skin diseases to exchange knowledge, update and develop best practice guidelines, and improve professional training and patient education.

It aims to improve healthcare organisation with the pooling of resources, including a platform with expert pathologists for a centralised study of slides and collaborative discussions on difficult cases. For every disease covered, core multidisciplinary teams will include a dermatologist, a nurse, a psychologist, a



A comprehensive socio-economic study on the individual burden of diseases will be conducted.

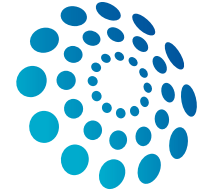
geneticist, a dietician and a pathologist, along with other specialists as required.

ERN Skin will also develop rare skin disease registries allowing participation in research programmes and clinical trials with well-characterised patients, as well as the stimulation of therapeutic research with sufficiently larger cohorts of patients. In addition, a comprehensive socio-economic study on the individual burden of diseases will be conducted. ■

NETWORK COORDINATOR

Professor Christine Bodemer
*Assistance Publique-Hôpitaux de Paris,
Hôpital Necker-Enfants Malades, France*

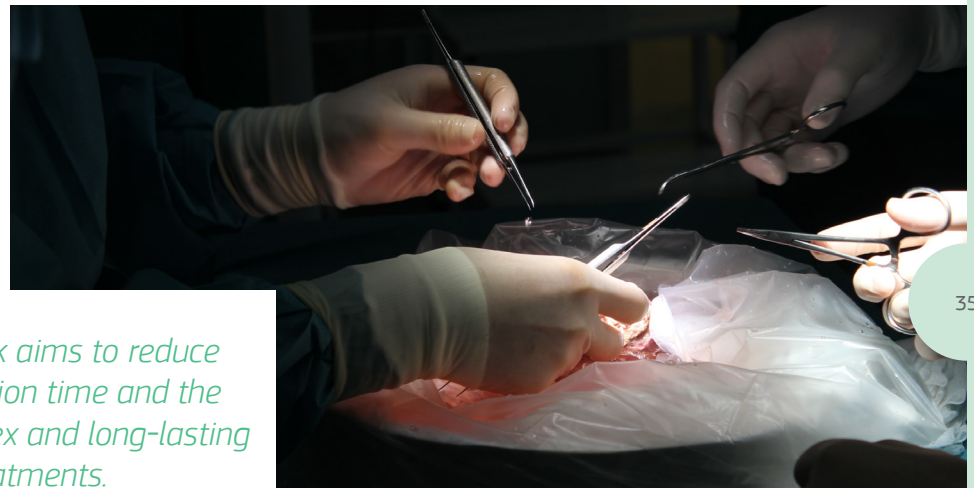
ERN on transplantation in children (ERN TRANSPLANT-CHILD)



Paediatric transplantation (PT), both solid organ (SOT) and haematopoietic stem cell transplantation (HSCT), is the only curative procedure for several rare diseases.

Optimal post-transplant care requires the concerted efforts of a multidisciplinary team. After transplantation, patients face chronic immunosuppression to avoid rejection. This requires monitoring for post-transplant complications to extend lifespans and improve quality of life. ERN TRANSPLANT-CHILD brings together experts in PT and post-transplantation care to improve outcomes for children and their families.

The network aims to reduce hospitalisation time and the use of complex and long-lasting treatments. It is working to improve psychological support services as children transition to adulthood. TRANSPLANT-CHILD aims to make available the latest techniques and



The network aims to reduce hospitalisation time and the use of complex and long-lasting treatments.

medical, pharmacological and therapeutic advances. Members are also facilitating the dissemination of harmonised clinical practice guidelines and the development of personalised medicine in PT.

TRANSPLANT-CHILD seeks to reduce the costs associated with transplantation — such as re-transplantation and pharmacological treatments — and is harmonising the PT care to minimise the risks of post-transplantation complications. Together, Europe's leading

PT experts are working to reduce mortality and morbidity related to transplantation in children.

NETWORK COORDINATOR

Dr Paloma Jara Vega
Hospital Universitario La Paz, Spain

Focusing on improving patients' health outcomes: the role of patient organisations

ERNs are about patients. Patient organisations and, in particular, EURORDIS, have played an active role in the development of the networks for more than a decade, helping to ensure that the priorities will be to enhance clinical excellence and to improve patients' health outcomes and equitable access to quality care across Europe. EURORDIS is a non-governmental patient-driven alliance representing 733 rare disease patient organisations in 64 countries.

'We were there at the birth of the idea in the High-Level Working Group on Health Services and Medical Care, where ERNs were translated into the Cross-Border Healthcare Directive,' Mr Matt Bolz-Johnson, Healthcare and Research Director at EURORDIS recalls. 'We have walked the long road with Member States and the

European Commission, from the birth of the concept into legislation, through to the germination of ERNs into reality.'

As a consistent partner in nurturing the concept of ERNs, EURORDIS has ensured patient involvement throughout and developed a strong technical understanding of how patients' engagement in the networks can add real life value for patients.

'Networks have the potential to unlock tangible benefits for patients with rare and complex presentations of disease,' says Mr Bolz-Johnson. 'ERNs will break the isolation that rare disease communities face and make experts visible to patients across Europe, magnetising patient needs to the right experts quicker.'

'Networks have the potential to unlock tangible benefits for patients with rare and complex presentations of disease.'

One of the key benefits of ERNs for patients will be their capacity to accelerate diagnosis and reduce the number of undiagnosed or misdiagnosed patients. Mr Bolz-Johnson says the networks will 'erode the diagnostic odyssey'.

For many rare diseases, there are no currently available treatments. However, the culture of learning that ERNs promise to create will make them a hotbed of innovation. By devising simple outcome measures for specific diseases, it will open the door to faster identification

and adoption of optimal medical or surgical interventions. 'This will enhance best practice as ERN members learn from one another,' explains Mr Johnson. **'Experts will be able to share cases in real time through virtual meetings and review outcomes retrospectively to see what works best.'**

Patients have high hope that ERNs can make a real impact on their lives: 'We believe that thanks to sharing experience and expertise, we could make better use of existing and create new knowledge, to see significant improvements in outcomes of care for many rare diseases within years of ERNs being established,' Mr Bolz-Johnson says. **'Now it is time for ERNs to deliver.'** ■



'ERNs will break the isolation that rare disease communities face and make experts visible to patients across Europe, magnetising patient needs to the right experts quicker.'

ERN on hereditary metabolic disorders (MetabERN)

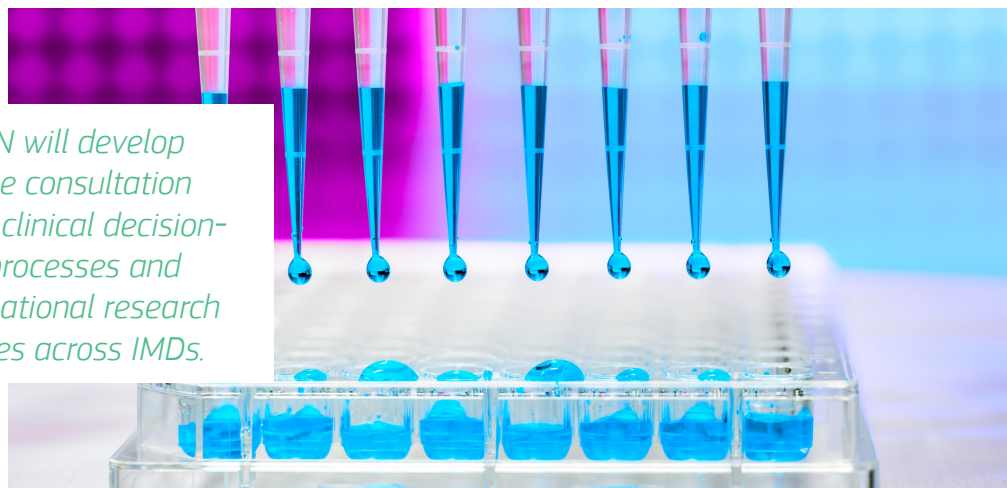


Rare inherited metabolic diseases (IMDs), of which there are more than 700, are individually rare but collectively frequent. Many metabolic diseases have severe, sometimes life-threatening, implications for patients. These conditions include disorders of all organs, can affect people of any age, and require multidisciplinary collaboration between a range of professionals.

Early diagnosis can improve outcomes but only 5 % of known IMDs are currently included in newborn screening programmes in Europe and there is a need for harmonisation of national programmes. For many of these conditions, knowledge about their natural history, the efficacy and safety of therapies, and long-term follow-up is incomplete.

MetabERN seeks to improve the lives of people affected by this highly heterogeneous group of diseases by dividing them into seven main

MetabERN will develop a real-time consultation platform for clinical decision-making processes and foster translational research programmes across IMDs.



categories. It is the first pan-European and pan-metabolic network of its kind.

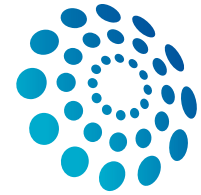
The network is setting up an inventory of metabolic diseases, developing patient information and training sessions, advancing collaborative diagnosis of new diseases, and establishing a long-term referral point bringing expertise to patients.

MetabERN will develop a real-time consultation platform for clinical decision-making processes and foster translational

research programmes across IMDs. It will share knowledge within the network and beyond by expanding to additional regions and countries.

NETWORK COORDINATOR

Professor Maurizio Scarpa
*Helios Dr Horst Schmidt Kliniken,
Germany*

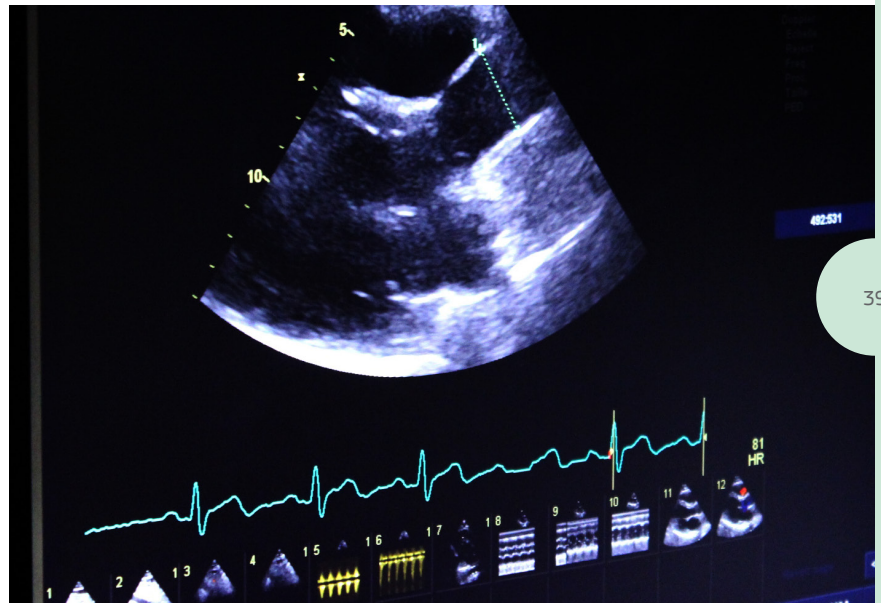


ERN on multisystemic vascular diseases (VASCERN)

Rare multisystemic vascular diseases include disorders which affect all types of blood vessels, with consequences for several bodily systems. These diseases require a multidisciplinary approach to care.

VASCERN comprises five Rare Diseases Working Groups: Hereditary Haemorrhagic Telangiectasia (HHT-WG), Heritable Thoracic Aortic Diseases (HTAD-WG), Medium Sized Arteries (vascular Ehlers Danlos Syndrome) (MSA-WG), Paediatric and Primary Lymphedema (PPL-WG) and Vascular Anomalies (VASCA-WG). A dedicated Patient-WG enables patient representatives to be involved in all activities of the ERN. In addition, several Thematic Working Groups are established to address communication, eHealth, ethics, patient registry, and training & education.

Networking, sharing and spreading expertise, promoting best practices, guidelines and clinical outcomes, patient empowerment, and improving knowledge through clinical and basic research are among VASCERN's objectives.



Health professionals involved in VASCERN will give lectures on their areas of expertise and make educational materials available online. One-week fellowships will be set up to allow EU students to learn more about these rare presentations, and knowledge will be shared through the network and with health professionals not covered by the ERN.

NETWORK COORDINATOR

Professor Guillaume Jondeau
*Assistance Publique-Hôpitaux de Paris,
Hôpital Bichat, France*

ERN Directory

Endo-ERN	European Reference Network on endocrine conditions
ERKNet	European Reference Network on kidney diseases
ERN BOND	European Reference Network on bone disorders
ERN CRANIO	European Reference Network on craniofacial anomalies and ENT disorders
ERN EpiCARE	European Reference Network on epilepsies
ERN EURACAN	European Reference Network on adult cancers (solid tumours)
ERN EuroBloodNet	European Reference Network on haematological diseases
ERN eUROGEN	European Reference Network on urogenital diseases and conditions
ERN EURO-NMD	European Reference Network on neuromuscular diseases
ERN EYE	European Reference Network on eye diseases
ERN GENTURIS	European Reference Network on genetic tumour risk syndromes
ERN GUARD-HEART	European Reference Network on diseases of the heart
ERNICA	European Reference Network on inherited and congenital anomalies
ERN ITHACA	European Reference Network on congenital malformations and rare intellectual disability
ERN LUNG	European Reference Network on respiratory diseases
ERN PaedCan	European Reference Network on paediatric cancer (haemato-oncology)
ERN RARE-LIVER	European Reference Network on hepatological diseases
ERN ReCONNET	European Reference Network on connective tissue and musculoskeletal diseases
ERN RITA	European Reference Network on immunodeficiency, autoinflammatory and autoimmune diseases
ERN-RND	European Reference Network on neurological diseases
ERN Skin	European Reference Network on skin disorders
ERN TRANSPLANT-CHILD	European Reference Network on transplantation in children
MetabERN	European Reference Network on hereditary metabolic disorders
VASCERN	European Reference Network on multisystemic vascular diseases

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Half a million people in Europe are diagnosed with a rare disease every year. No country can meet this challenge alone.

European Reference Networks are virtual networks that bring together experts from across the EU.

Together, they will tackle complex or rare diseases by improving diagnosis and access to specialist care.

More on European Reference Networks



<http://ec.europa.eu/health/ern/>

