European Reference Network
for rare or low prevalence complex diseases

European Commission

ERN, CPMS a spol:
Co by měl pacient vědět....

Pavla Doležalová

Centrum dětské revmatologie a autoinflamatorních onemocnění

Klinika pediatrie a dědičných poruch metabolismu 1.LFUK a VFN v Praze

European Reference Network
for rare or low prevalence complex diseases

Network
Immunodeficiency, Autoinflammatory and Autoimmune Diseases
(ERN RITA)

Rare Immunodeficiency auToinflammatory Autoimmune

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Příběh Violety

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Příběh Violety

Rare Immunodeficiency auToinflammatory Autoimmune
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- 3 měsíce:
  - Horečky, lokalizovaný edém (úbouleů) krvavý průjem, vysoká zánětlivá aktivita
- Vyloučení infekce, malignit, PID
- U nás v 6 měsících života
  - autoinflamatorní/systémové onemocnění ?
- Anakinra 10 mg/kg
  - Ústup horeček a průjmu

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Příběh Violety pokračuje

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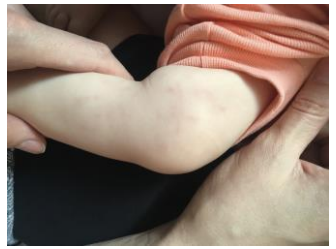
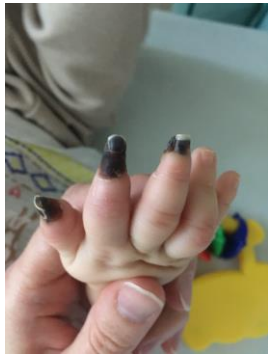
- Během 2 týdnů:
  - Livedo a ischemie prstů
- DADA2 ? Jiná monogenní vaskulitida?
  - AI panel ….čekáme na výsledek
  - Prostacyclin, pulzy SM, prednison, etanercept
  - Po 2 měsících mírný pokles zánětlivých parametrů, zastavena progrese digitální ischemie
- ADA2 normální
  - Co když nedokážeme vysadit KS??
  - CO MÁM DĚLAT?





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Co dál?

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- ~~Panikařím~~
- Volám příteli
- Píšu email do amerického diskuzního klubu ped-rhe Bulletin Board

  - Může to být DADA2 když je ADA2 v normě?
  - Měříme ADA2 správně?
  - Udělá nám někdo genetiku urychleně?
  - Jaká další vyšetření mám udělat?
  - Může 3-měsíční kojenec mít skutečnou PAN?
  - Co jiného to může být?
  - Jaké další biologikum má dostat?

  ……

- Modlím se ….. a…..

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Co dělat, když jsem už opravdu v koncích?

Rare Immunodeficiency auToinflammatory Autoimmune

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Kontaktuj nejbližší ERN (RITA) zařízení

- Ve vlastní zemi
- V sousední zemi

Zažádej o mezinárodní expertní konzílium v systému CPMS

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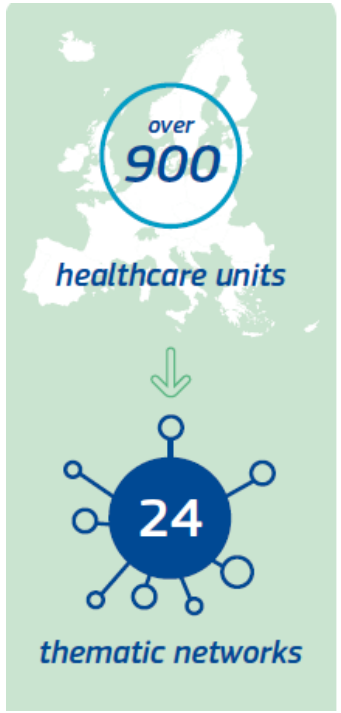
What are ERNs?

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.



No country alone has the knowledge and capacity to treat all rare and complex diseases.

Co jsou ERN?

European Reference Network
for rare or low prevalence complex diseases

Network
Immunodeficiency, Autoinflammatory and Autoimmune Diseases (ERN RITA)

Member
Všeobecná fakultní nemocnice v Praze — Česká republika

Hlavní cíle ERN

- Sdílení vědomostí a expertízy
- Pomoc s diagnostikou a léčbou
- Zajištění dostupnosti léčebných postupů

Cesty k jejich dosažení

- Virtuální sítě nemocnic
  - Tvorba virtuálních poradních sborů
- Vývoj inovativních modelů péče a nástrojů eHealth
- Rozvoj přeshraniční péče
- Možnosti financování ze strany EU

European Reference Network
for rare or low prevalence complex diseases

Co je CPMS ?

Rare Immunodeficiency auToinflammatory Autoimmune
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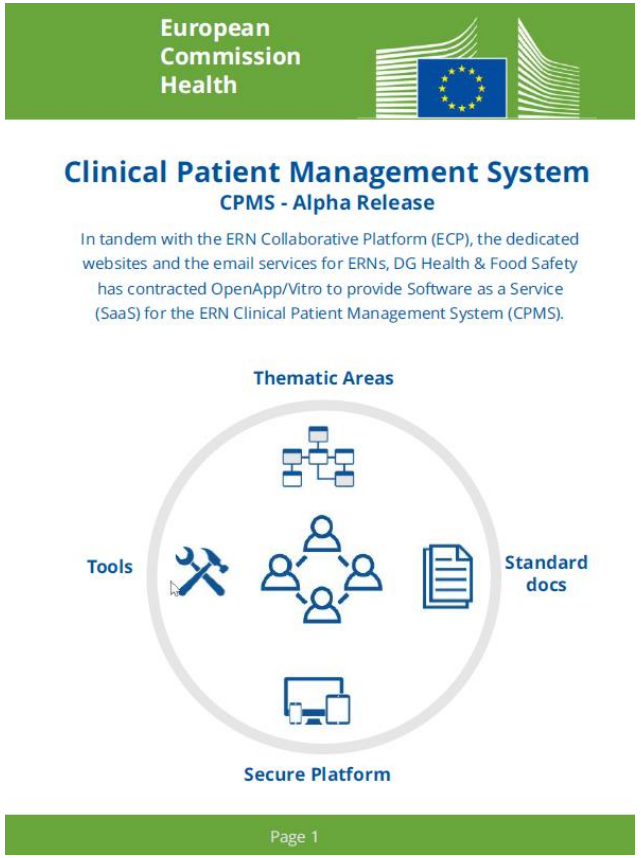
- Spolupráce a sdílení
  - Kazuistika sdílená mezi expertními pracovišti prostřednictvím zabezpečené online platformy
- Technické zázemí poskytnuto EK
  - User ID a heslo – dvojité zabezpečení
  - Pseudonymizace pacientů



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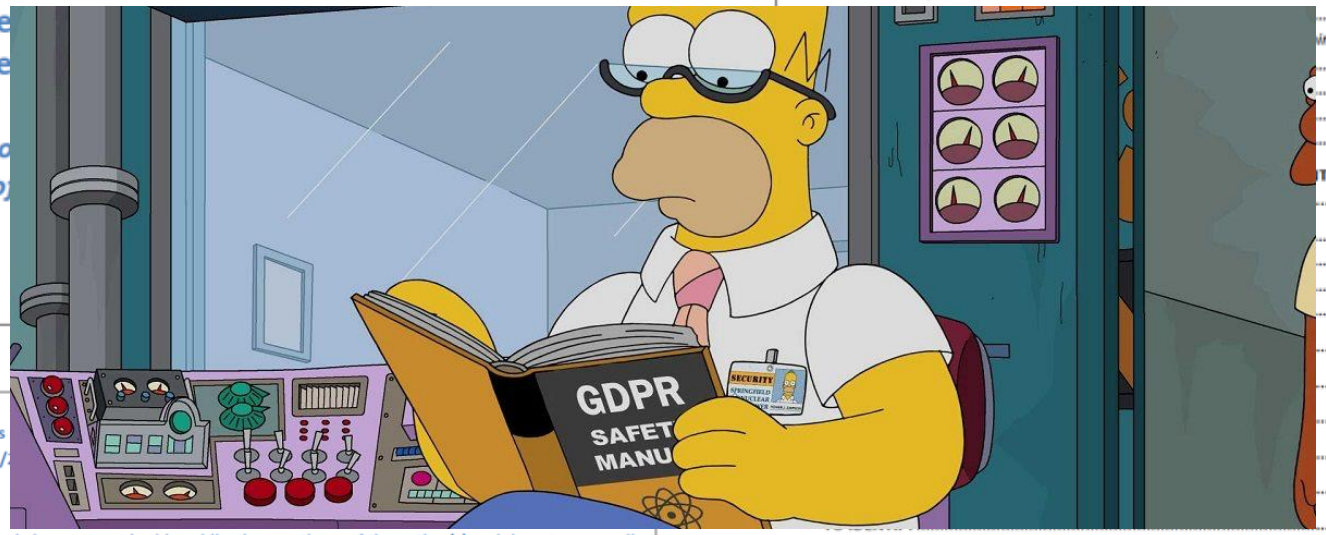
Ochrana osobních údajů

Rare Immunodeficiency auToinflammatory Autoimmune

European Reference Network

Good Practice
Europe

An overview of goo
context o



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Algoritmus konzultace

Rare Immunodeficiency auToinflammatory Autoimmune

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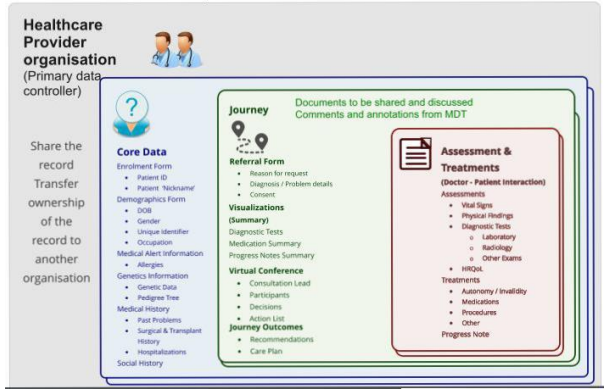
  - Diagnóza
  - Terapie
- Ošetřující lékař
  - Poskytne klinické informace
  - Může být členem konzultačního týmu (panelu)
- Vedoucí panelu
  - Vybírá členy
  - Svolává virtuální multidisciplinární konzultaci
  - Zaznamená výstup – závěr konzultace s doporučením



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CPMS funkcionality

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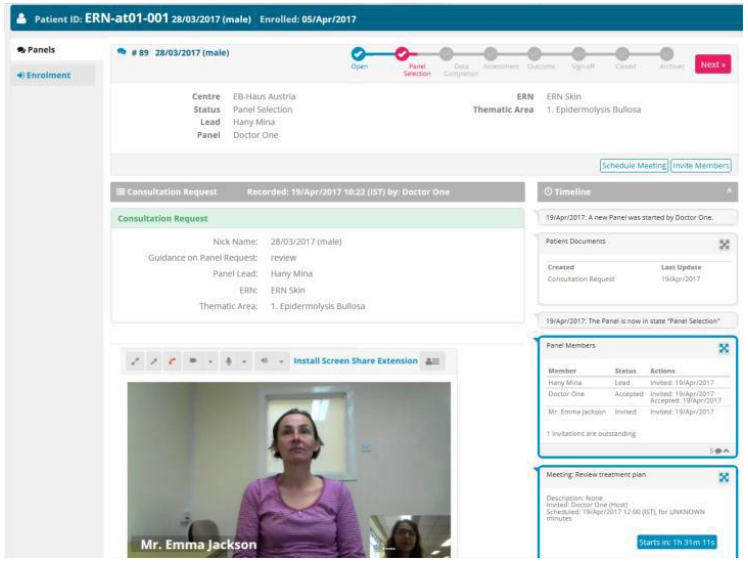

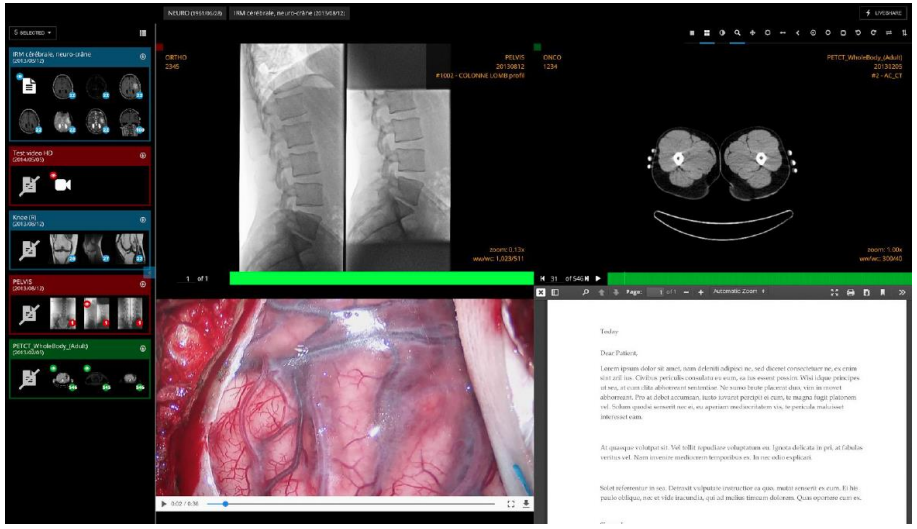

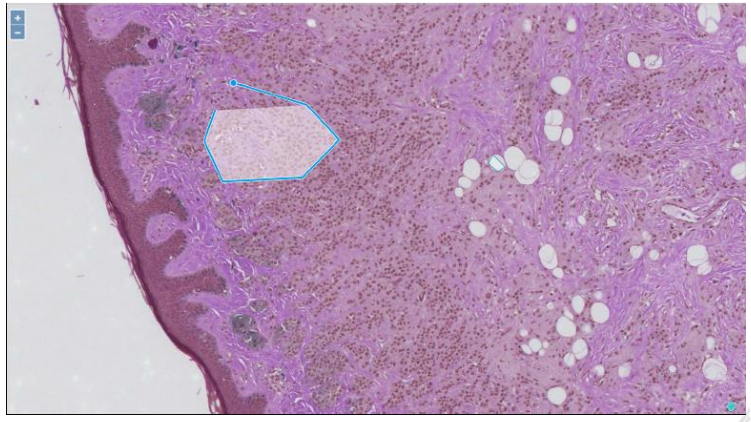

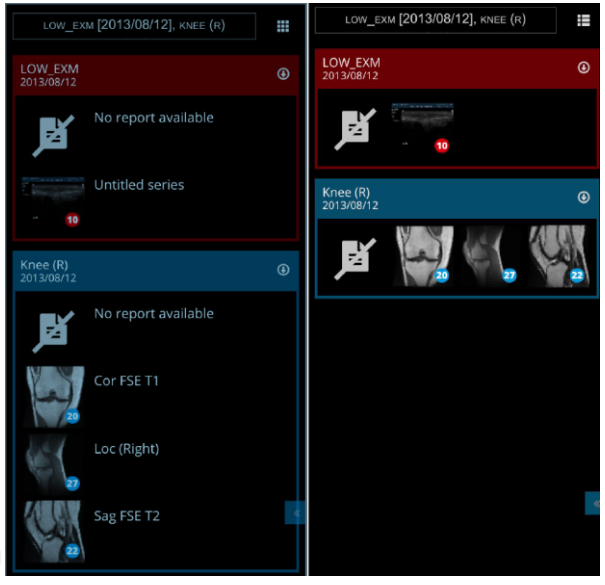

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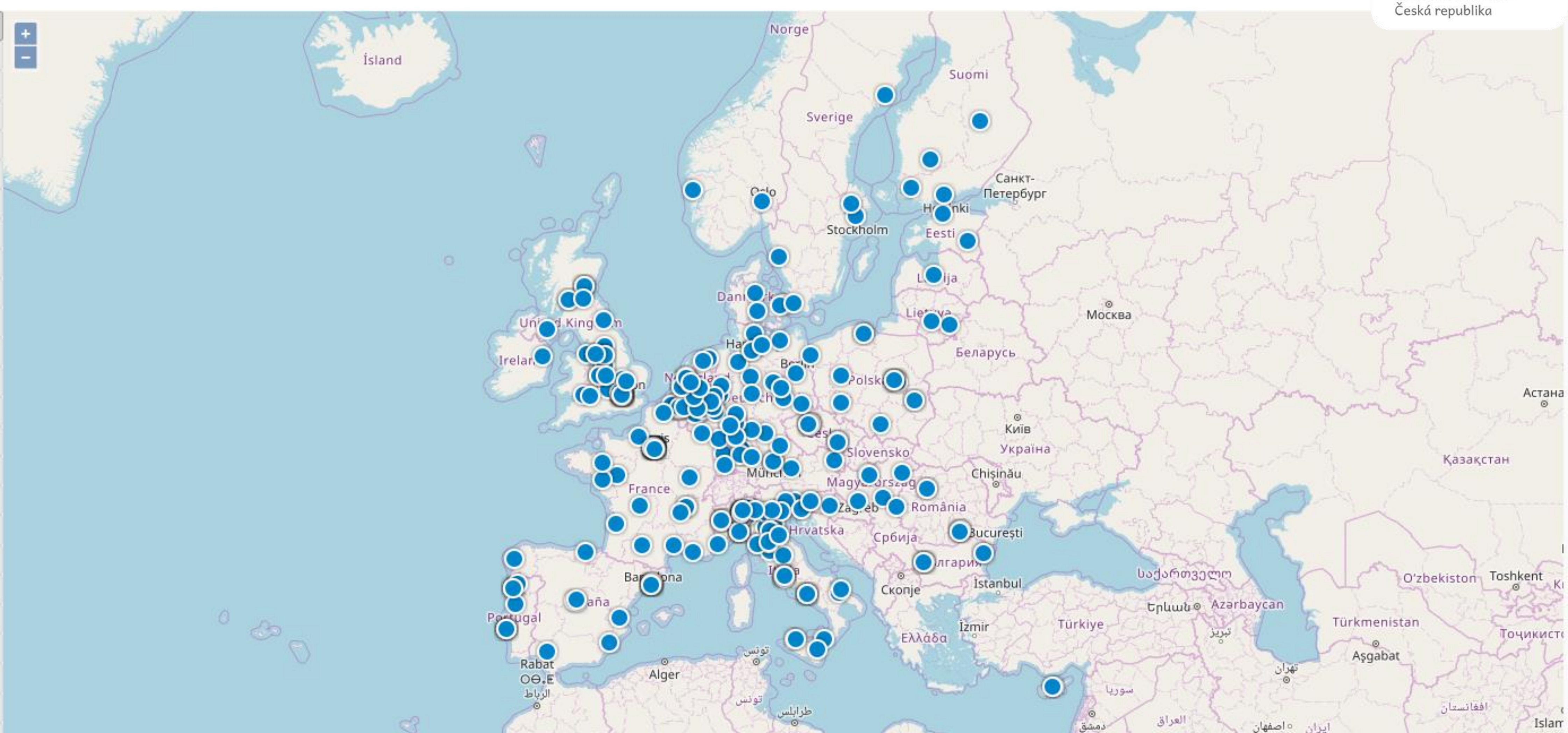
ERN celkově

European Reference Network
for rare or low prevalence complex diseases

Network
Immunodeficiency, Autoinflammatory and Autoimmune Diseases
(ERN RITA)

Member
Všeobecná fakultní nemocnice v Praze — Česká republika

- All HCPs
- ERN BOND
- ERN CRANIO
- Endo-ERN
- ERN EpiCARE
- ERKNet
- ERN RARE-LIVER
- ERN-RND
- ERN EYE
- ERNICA
- ERN LUNG
- ERN Skin
- ERN EURACAN
- ERN EURO-NMD
- ERN EuroBloodNet
- ERN eUROGEN
- ERN GENTURIS
- ERN GUARD-HEART
- ERN ITHACA
- MetabERN
- ERN PaedCan
- ERN ReCONNECT
- ERN RITA
- ERN TRANSPLANT-CHILD



ERN v ČR

ERN	HCP
GUARD-HEART	FN Motol Praha
ERN LUNG	FN Motol Praha
	VFN Praha
	Thomayerova nemocnice Praha
Endo-ERN	FN Motol Praha
	FN Královské Vinohrady Praha
ERN BOND	FN Motol Praha
ERKNet	FN Motol Praha
EURACAN	FN Motol Praha
	Masarykův onkologický ústav Brno
PaedCan-ERN	FN Motol Praha
	Fakultní nemocnice Brno
ITHACA	FN Motol Praha
EURO-NMD	FN Motol Praha
	Fakultní nemocnice Brno
EpiCARE	FN Motol Praha
	Fakultní nemocnice u sv. Anny v Brně
	Fakultní nemocnice Brno
CRANIO	FN Motol Praha
ERN-Skin	Nemocnice na Bulovce
	Fakultní nemocnice Královské Vinohrady
	Fakultní nemocnice Brno
	Fakultní nemocnice u sv. Anny v Brně
MetabERN	VFN Praha
ERN-RND	VFN Praha
ERN-EYE	VFN Praha
ERN RITA	VFN Praha
EuroBloodNet	Fakultní nemocnice Brno

European Reference Network
for rare or low prevalence complex diseases

Network
Immunodeficiency, Autoinflammatory and Autoimmune Diseases (ERN RITA)

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http://ec.europa.eu/health/ern/networks_en

FN Motol Praha

FN Královské Vinohrady Praha (VFN Praha)

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

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 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

FN Motol Praha
(IKEM Praha)

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.

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

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 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
Universitätsklinikum Heidelberg, Germany

FN Motol Praha

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.

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 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 Sangiori
Rizzoli Orthopaedic Institute, Bologna, Italy

FN Motol Praha

ERN on craniofacial anomalies and ENT disorders (ERN CRANIO)

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.

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

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 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

FN Motol Praha
Fakultní nemocnice u sv. Anny v Brně
Fakultní nemocnice Brno

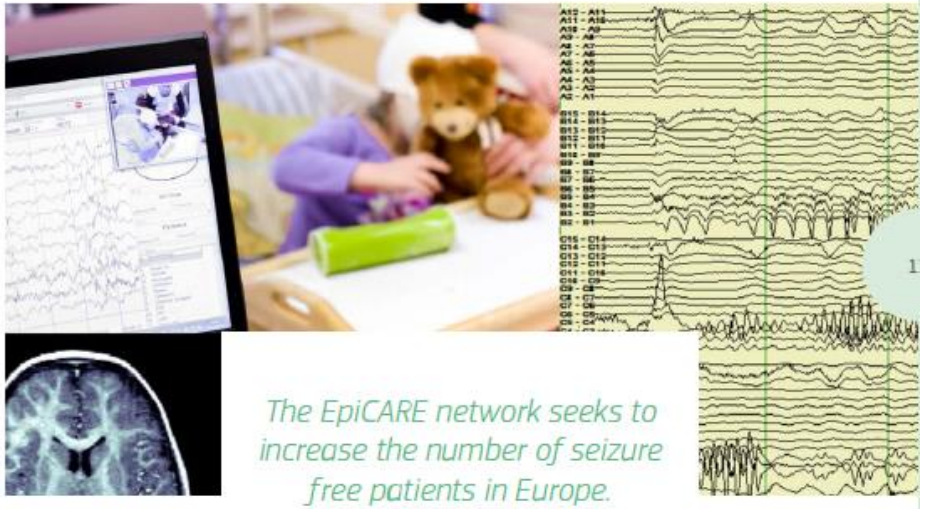
ERN on epilepsies (EpiCARE)

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-pilepsy which worked to increase awareness and accessibility of epilepsy surgery, for carefully selected individuals, that effectively 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.



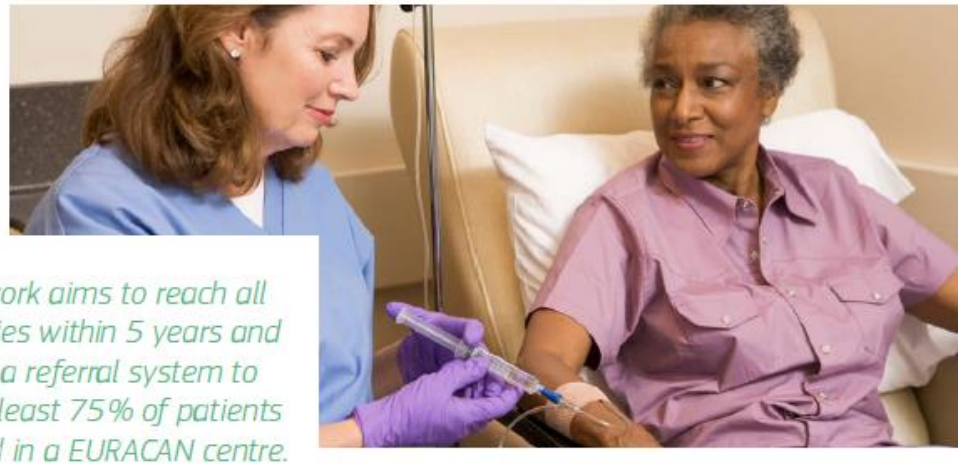
The EpiCARE network seeks to increase the number of seizure free patients in Europe.

NETWORK COORDINATOR

Professor Helen Cross
Great Ormond Street Hospital for Children NHS Foundation Trust, United Kingdom

FN Motol Praha
Masarykův onkologický ústav Brno
(Thomayerova nemocnice Praha)
(ÚPMD Praha)

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

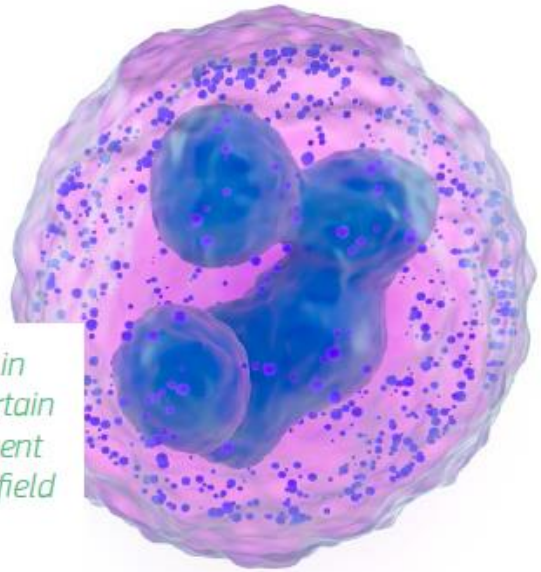
Professor Jean-Yves Blay
Centre Léon Bérard, Lyon, France

Fakultní nemocnice Brno

(ÚHKT Praha)

(FN Olomouc)

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.

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

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.

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

FN Motol Praha
Fakultní nemocnice Brno

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.

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.

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.

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

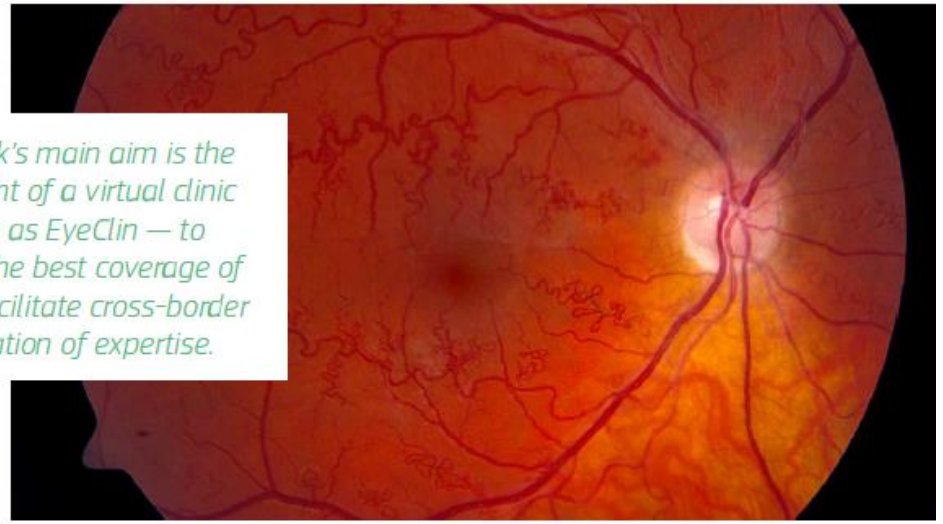
Professor Kate Bushby
The Newcastle upon Tyne Hospitals NHS Foundation Trust, United Kingdom

VFN Praha

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.

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.



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.

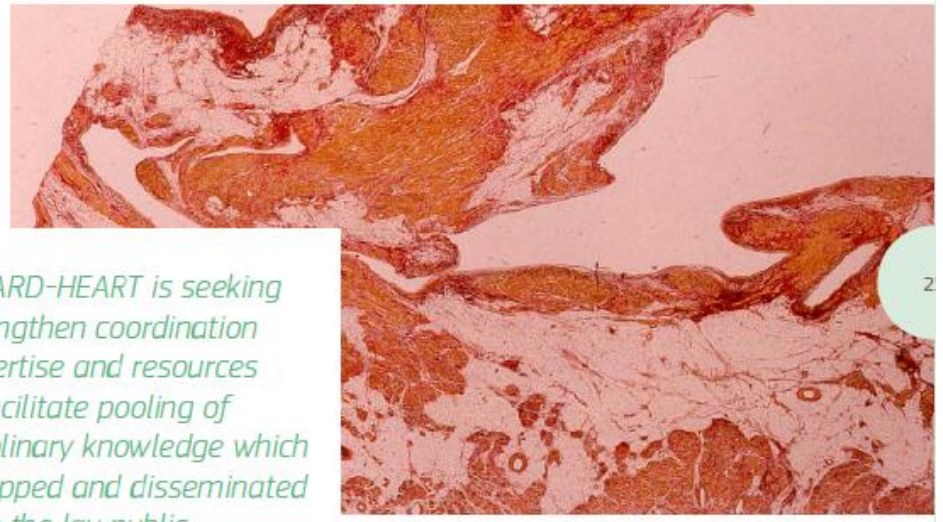
NETWORK COORDINATOR

Professor Hélène Dollfus
Hôpitaux Universitaires de Strasbourg, France

FN Motol Praha
(IKEM Praha)

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.



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.

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.

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

FN Motol Praha

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

Professor Jill Clayton-Smith
Central Manchester University Hospitals NHS Foundation Trust, United Kingdom

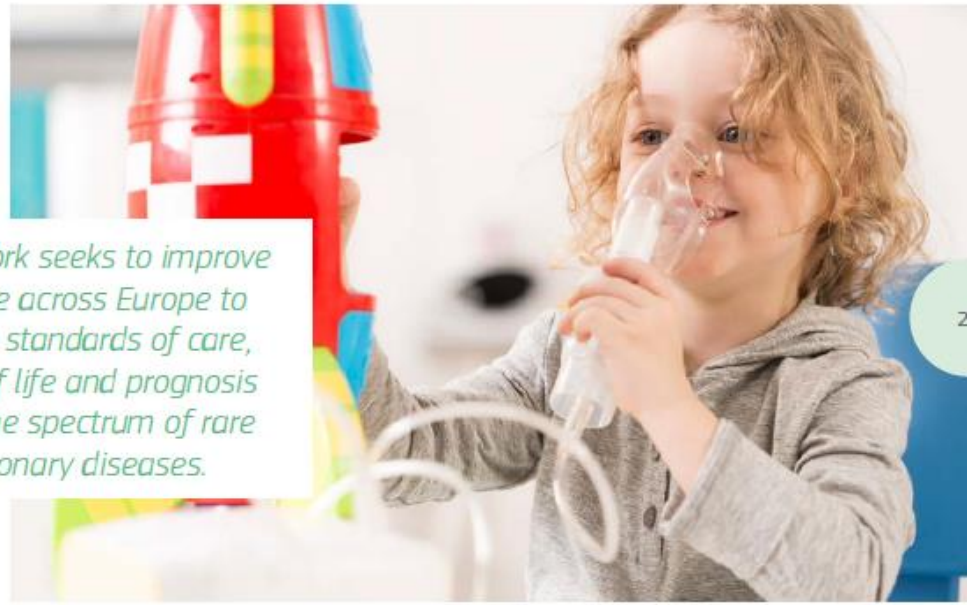
FN Motol Praha
VFN Praha
Thomayerova nemocnice Praha
(FN Brno)

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



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.

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

FN Motol Praha
Fakultní nemocnice Brno

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.

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

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 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 Kinderkrebsforschung, Austria

VFN Praha
(Thomayerova nemocnice Praha)
(FNUSA Brno)

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.

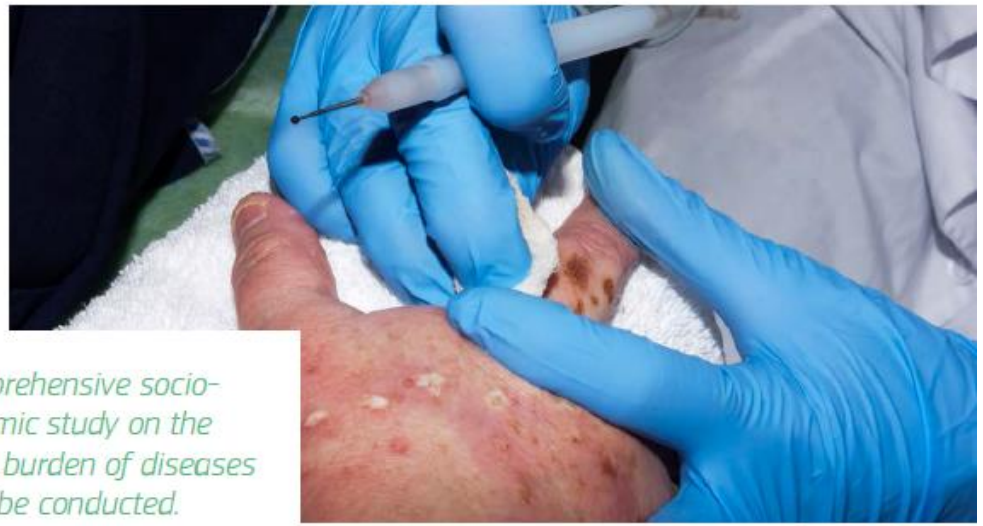
NETWORK COORDINATOR

Dr Holm Graessner
Universitätsklinikum Tübingen, Germany

Nemocnice na Bulovce
Fakultní nemocnice Královské Vinohrady
Fakultní nemocnice Brno
Fakultní nemocnice u sv. Anny v Brně

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.



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

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

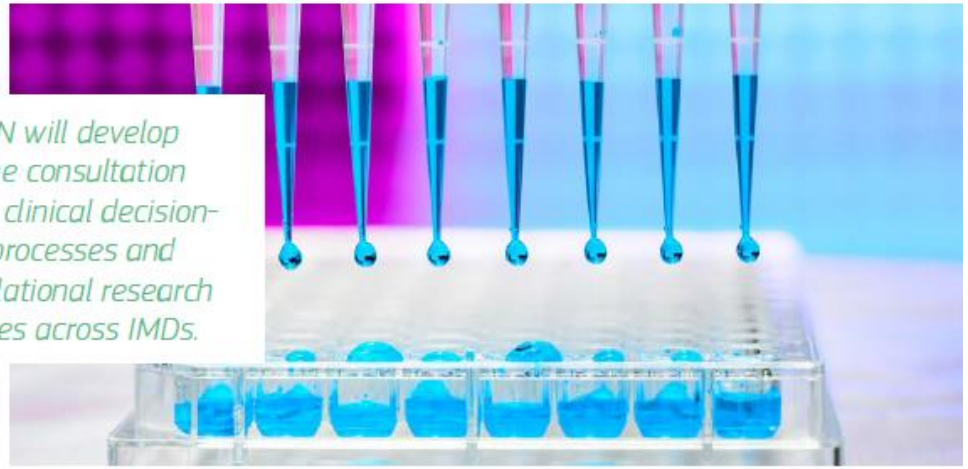
ERN Skin will also develop rare skin disease registries allowing participation in research programmes and clinical trials with well-characterised patients, as well as the stimulation of therapeutic research with sufficiently larger cohorts of patients. In addition, a comprehensive socio-economic study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

NETWORK COORDINATOR

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

VFN Praha

ERN on hereditary metabolic disorders (MetabERN)



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

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 heterogenous group of diseases by dividing them into seven main 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.

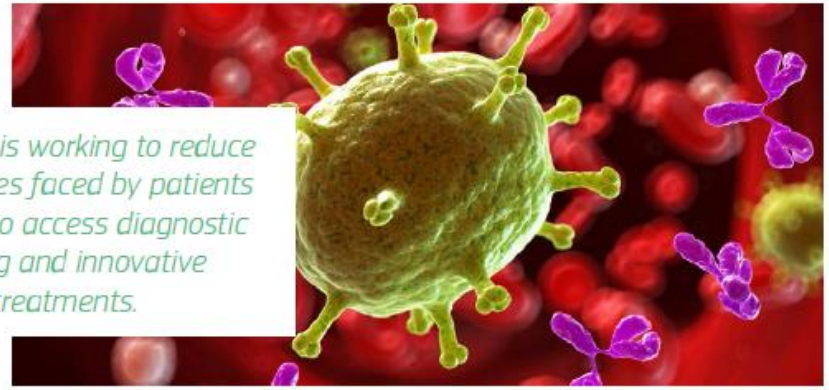
MetabERN will develop a real-time consultation platform for clinical decision-making processes and foster translational research programmes across IMDs. It will share knowledge within the network and beyond by expanding to additional regions and countries.

NETWORK COORDINATOR

Professor Maurizio Scarpa
Helios Dr Horst Schmidt Kliniken, Germany

VFN Praha

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



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

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 autoinflammatory 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 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

Professor Andrew Cant
The Newcastle upon Tyne Hospitals NHS Foundation Trust, United Kingdom

Žadatelé o členství v ERN

Fakultní nemocnice v Motole
European Reference Network on inherited and congenital anomalies (ERNICA)
Evropská referenční síť pro vzácné vrozené vady
European Reference Network on genetic tumour risk syndromes (GENTURIS)
Evropská referenční síť pro syndromy s rizikem nádorového onemocnění
Rare Immunodeficiency, Autoinflammatory and Autoimmune Diseases Network (RITA)
Evropská referenční síť pro vzácná imunodeficitní, autoinflamatorní a autoimunní onemocnění
Všeobecná fakultní nemocnice
European Reference Network on Rare Endocrine Conditions (Endo-ERN)
Evropská referenční síť pro vzácná endokrinologická onemocnění
European Reference Network on urogenital diseases and conditions (eUROGEN)
Evropská referenční síť pro vzácná urogenitální onemocnění
Institut klinické a experimentální medicíny
European Rare Kidney Diseases Reference Network (ERKNet)
Evropská referenční síť pro vzácná onemocnění ledvin
Gateway to Uncommon And Rare Diseases of the HEART (GUARD-HEART)
Evropská referenční síť pro vzácná onemocnění srdce
European Reference Network on hepatological diseases (RARE-LIVER)
Evropská referenční síť pro vzácná onemocnění jater

Thomayerova nemocnice
European Reference Network on urogenital diseases and conditions (eUROGEN)
Evropská referenční síť pro vzácná urogenitální onemocnění
European Reference Network on Rare Adult Cancers (solid tumors) (EURACAN)
Evropská referenční síť pro vzácná nádorová onemocnění dospělých
European Reference Network on Rare Neurological Diseases (ERN-RND)
Evropská referenční síť pro vzácná neurologická onemocnění
Revmatologický ústav
European Reference Network on connective tissue and musculoskeletal diseases (ReCONNET)
Evropská referenční síť pro vzácná onemocnění pojivové tkáně a pohybového aparátu
Ústav pro péči matku a dítě
European Reference Network on Rare Adult Cancers (solid tumors) (EURACAN)
Evropská referenční síť pro vzácná nádorová onemocnění dospělých
Ústav hematologie a krevní transfuze
European Reference Network on Rare Hematological Diseases (EuroBloodNet)
Evropská referenční síť pro vzácná hematologická onemocnění

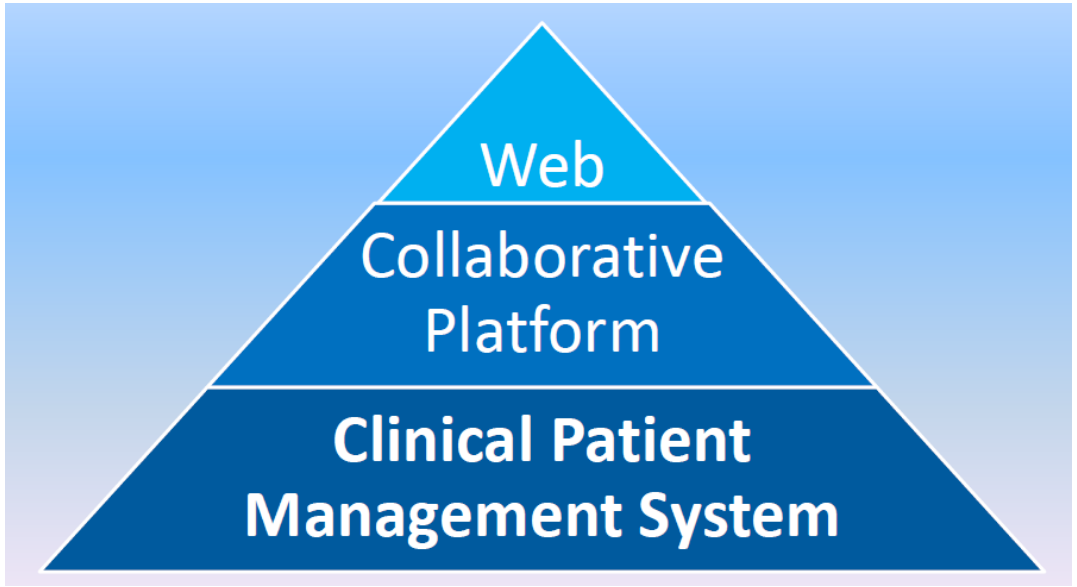
Fakultní nemocnice u sv. Anny v Brně
European Reference Network on Rare Neurological Diseases (ERN-RND)
Evropská referenční síť pro vzácná neurologická onemocnění
Fakultní nemocnice Brno
European Reference Network on Rare Respiratory Diseases (ERN-LUNG)
Evropská referenční síť pro vzácná respiračních onemocnění
Fakultní nemocnice Olomouc
European Reference Network on Rare Hematological Diseases (EuroBloodNet)
Evropská referenční síť pro vzácná hematologická onemocnění
Masarykův onkologický ústav
European Reference Network on genetic tumour risk syndromes (GENTURIS)
Evropská referenční síť pro syndromy s rizikem nádorového onemocnění

European Reference Network
for rare or low prevalence complex diseases

Network
Immunodeficiency, Autoinflammatory and Autoimmune Diseases (ERN RITA)

Member
Všeobecná fakultní nemocnice v Praze — Česká republika

Struktura ERN: Pracovní skupiny



http://rita.ern-net.eu

https://webgate.ec.europa.eu/ern/

Kolaborativní platforma

European Reference Network
for rare or low prevalence complex diseases

Network
Immunodeficiency, Autoinflammatory and Autoimmune Diseases (ERN RITA)

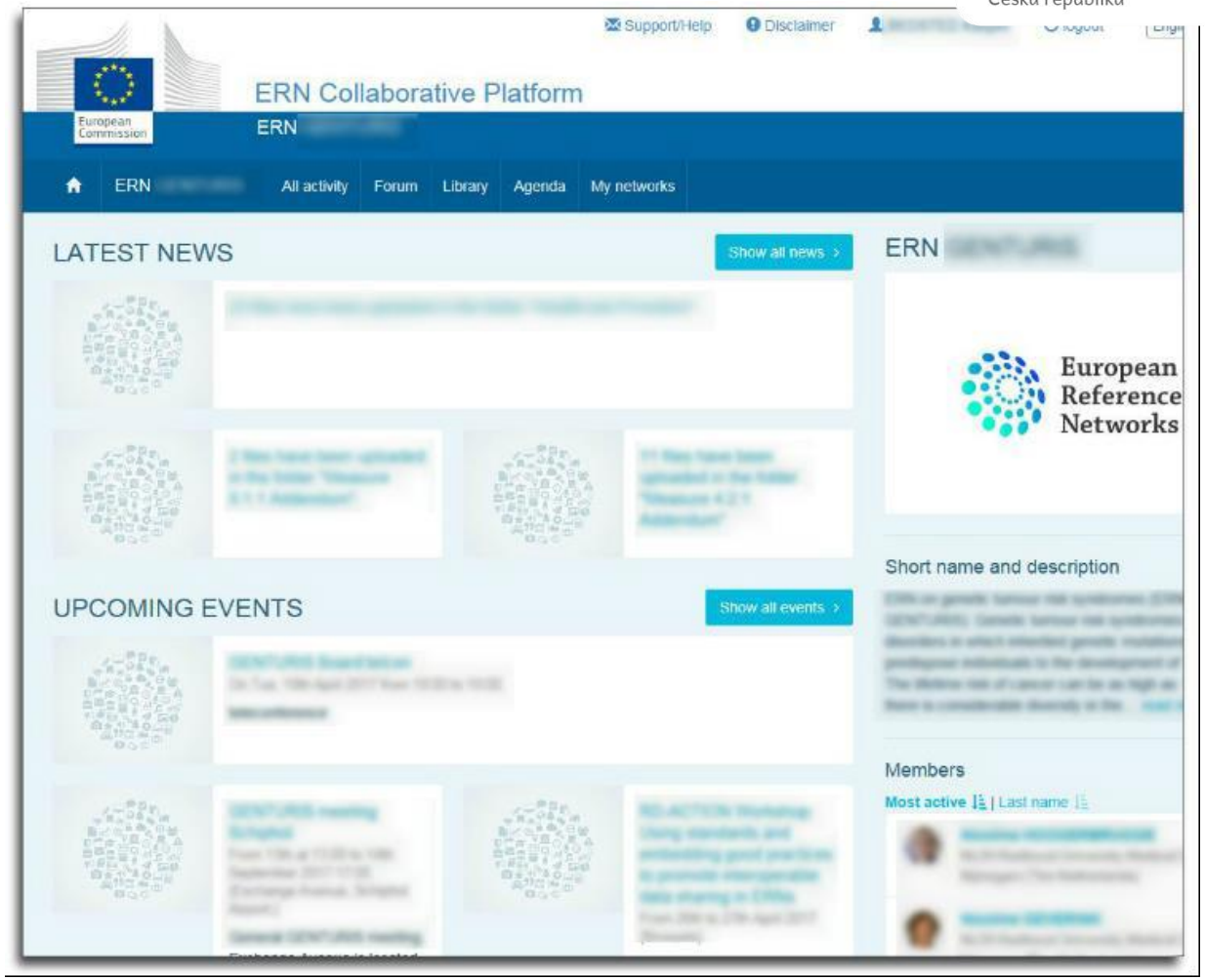
Member
Všeobecná fakultní nemocnice v Praze — Česká republika

- **Účel – podpora spolupráce:**
  - **Online komunikace**
  - **Sdílení dokumentů**
  - **Organizace akcí**
  - **Neslouží pro klinické informace**

- **EC Demo Video:**

https://webgate.ec.europa.eu/ern/imgs/ECP overview video.mp4

- *SANTE-ERN-ECP-ITSUPPORT@ec.europa.eu*

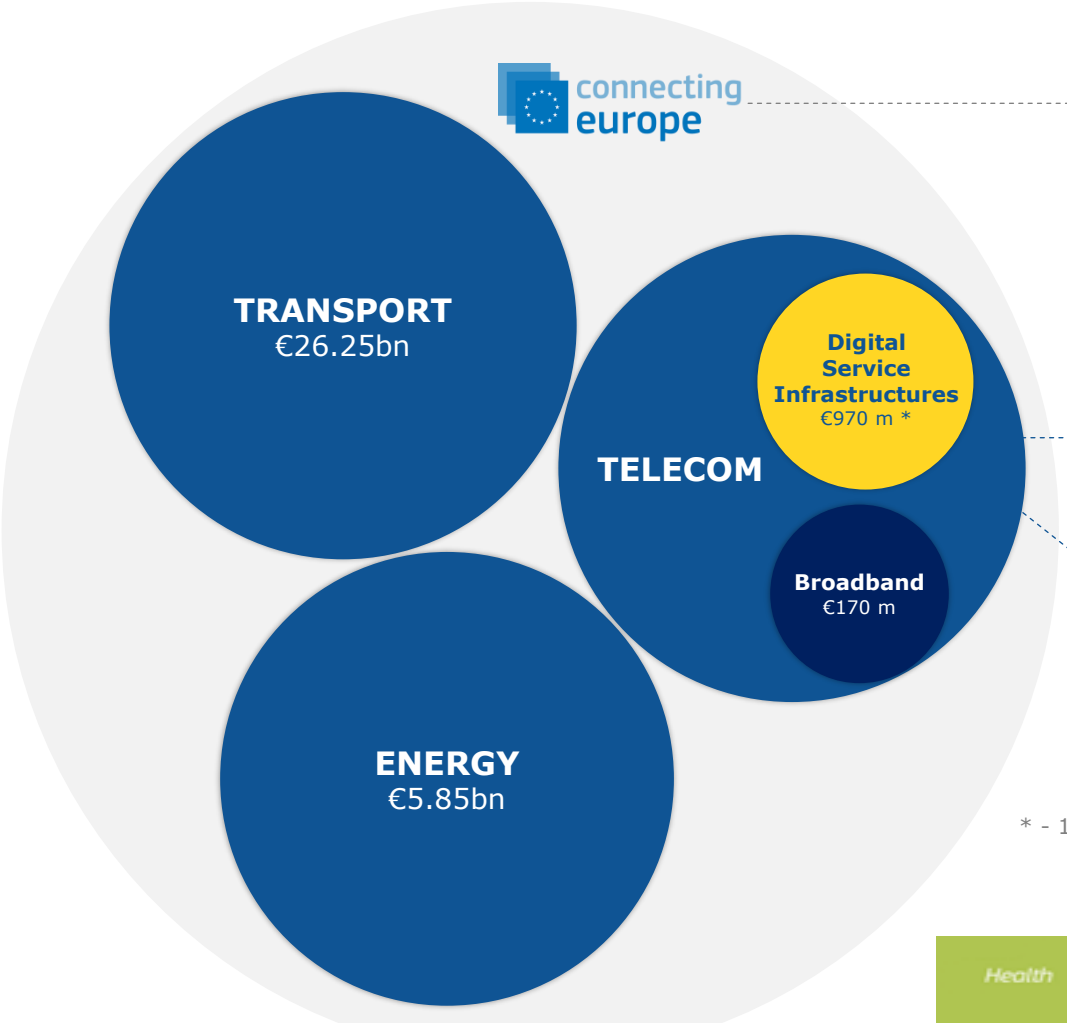


CPMS podpora

- Koordinátorská IT advisory group
- ERN IT working group
- Individuální IT/eHealth pracovní skupina při každé ERN

- „Helpdesk“
  - „Core“ – pro CP a CPMS, provozovaná EC – DG Sante
  - Individuální pro jednotlivé ERNy – předmět grantové aplikace

Connecting Europe Facility



HOW IS IT REGULATED?

CEF Regulation (REGULATION (EU) No 1316/2013)
The Connecting Europe Facility (CEF) is a regulation that defines how the Commission can finance support for the establishment of trans-European networks to reinforce an interconnected Europe.

CEF Work Programme
Translates the CEF Telecom Guidelines in general objectives and actions planned on a yearly basis.

****DSI** describe solutions that support the implementation of EU-wide projects. They provide trans-European interoperable services and which are composed of core service platforms and generic services.

* - 100 m Juncker Package

Health

CEF Telecom – what does it finance

DIGITAL SERVICE INFRASTRUCTURES (DSIs)

EUROPEAN COMMISSION

CORE SERVICE PLATFORM
(Offered by the European Commission:
Clinical Patient Management System
ERN Collaborative Platform)

ERN

GENERIC SERVICES
(Grants for projects for ERN)

Health

eHealth DSI Call for ERN

- Timeline: 6 May – 21 September 2017
- Budget: ~€3 million; proposals requesting a contribution of up to €125,000 are expected
- Indicative duration: 12 months
- Co-funding rate: 75%

European Reference Network
for rare or low prevalence complex diseases

Zdravotní péče o pacienty s VO

European Commission

- Potřeba komplexní, často multidisciplinární péče
  - Příslušnost k definovaným specializačním oborům nejasná, na okraji zájmu odborných společností
- Síť specializovaných pracovišť
  - Není zatím pro většinu VO oficiálně ustanovena
  - Expertní pracoviště (evropská referenční centra, ERN) čekají na přidělení statusu Center vysoce specializované péče
  - Kapacita center omezená, na okraji zájmu poskytovatelů pro ekonomickou nevýhodnost
- Formulace potřebných změn v organizaci a financování péče o pacienty s VO
  - Podmínkou je znalost počtu a rozložení jednotlivých skupin VO

pavla.dolezalova@vfn.cz

European Reference Network
for rare or low prevalence complex diseases

Data o VO = základní předpoklad pro změny

European Commission

- **Informace** o
  - počtech pacientů
  - rozložení diagnóz
  - nákladech na péči
- **nejsou v ČR k dispozici**
  - Pacienty nelze v nemocničních EHR identifikovat
  - Diagnózy dle MKN vesměs nepřesné

pavla.dolezalova@vfn.cz

European Reference Network
for rare or low prevalence complex diseases

Systémové kroky v péči o pacienty s VO

European Commission

- Projekt mapování nákladovosti péče o pacienty s VO s cílem optimalizace úhrad
  - Spolupráce pilotních poskytovatelů s plátci (VZP) a ÚZIS
  - Identifikace pacientů s VO na 5 pilotních pracovištích 3 poskytovatelů pomocí „signálního kódu“
  - Poskytne demografická data + nákladovost bez vazby na přesnou Dg
- Projekt „národního registru VO“ (RARE)
  - Projekt ÚZIS ve spolupráci s odborníky
  - Identifikace obecného datasetu zdravotních dat (CDE) ve standartní struktuře, interoperabilní s mezinárodními registry
  - Základní parametr CDE = diagnóza VO pomocí Orpha-kódu

pavla.dolezalova@vfn.cz

„Pilotní projekt“ – první fáze

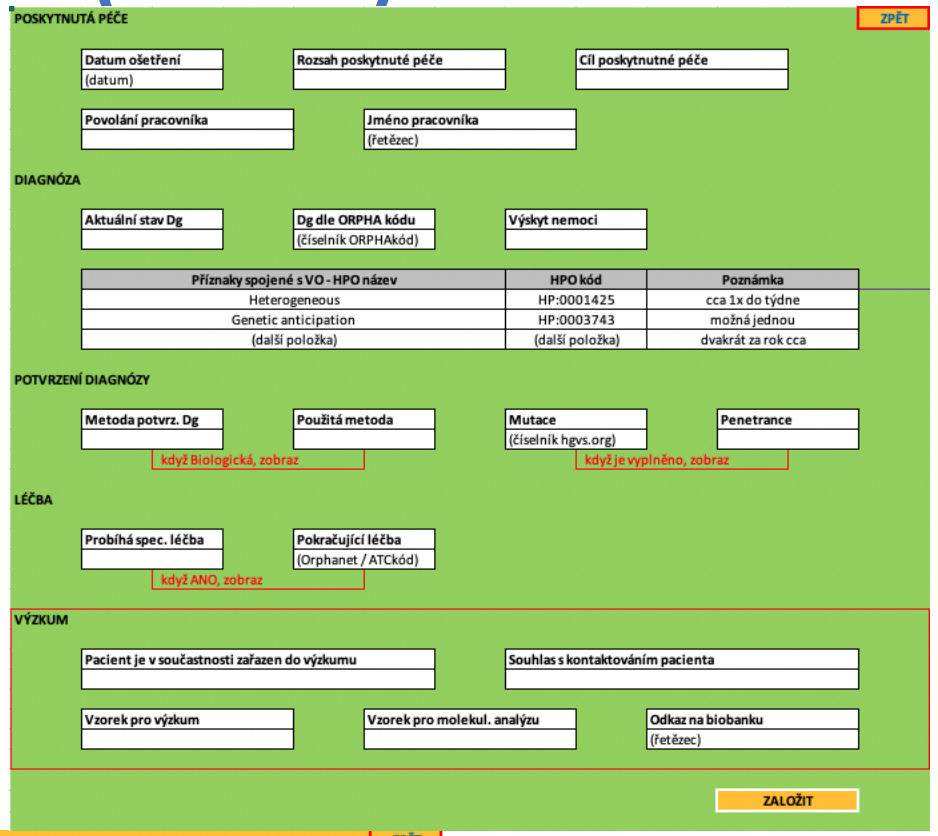
- Projekt mapování nákladovosti péče o pacienty s VO s cílem optimalizace úhrad
  - Spolupráce pilotních poskytovatelů (FNM, VFN, FNB) s plátci (VZP) a ÚZIS
  - Identifikace pacientů s VO na 5 pilotních pracovištích 3 poskytovatelů pomocí „signálního kódu“
    - FNM: ERN Lung, ERN-NMD
    - VFN: Metab ERN, ERN RITA
    - FNB: Euro Blood Net
  - Poskytne demografická data + nákladovost bez vazby na přesnou Dg
- Zadání analýzy 1.roku projektu (1.1.-31.12.2020)
  - **Identifikace pacientů s VO**: Signální kód 99976 + kód MKN10
  - počet vyšetřených pacientů, jejich věk a diagnózy (MKN)
  - počet a typ výkonů, celková úhrada / rok, průměrná úhrada na pacienta / rok
  - vykázaná pomocná vyšetření (laboratorní, zobrazovací, funkční atd) provedená daným poskytovatelem
  - vyžádaná péče a pomocná vyšetření provedená mimo poskytovatele „centrové“ péče / rok
  - léčivé přípravky – celková suma / rok a průměr na pacienta
  - poměr mezi péčí poskytnutou centrovým poskytovatelem a jinými zařízeními
  - identifikace počtu pacientů, kteří neprošli v daném roce centrem a byli sledování jinde + přehled těchto pracovišť

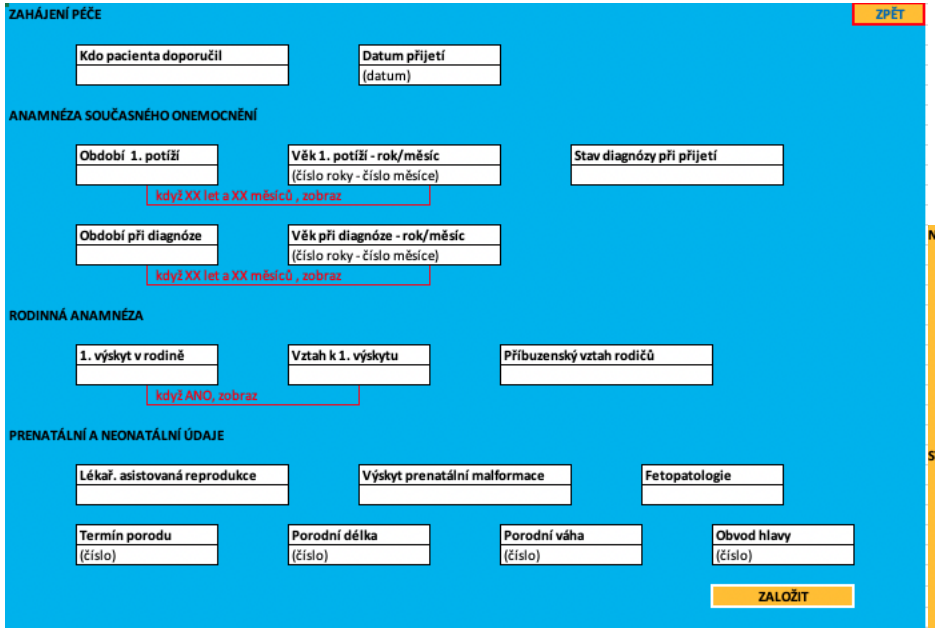
„Pilotní projekt“ - pokračování

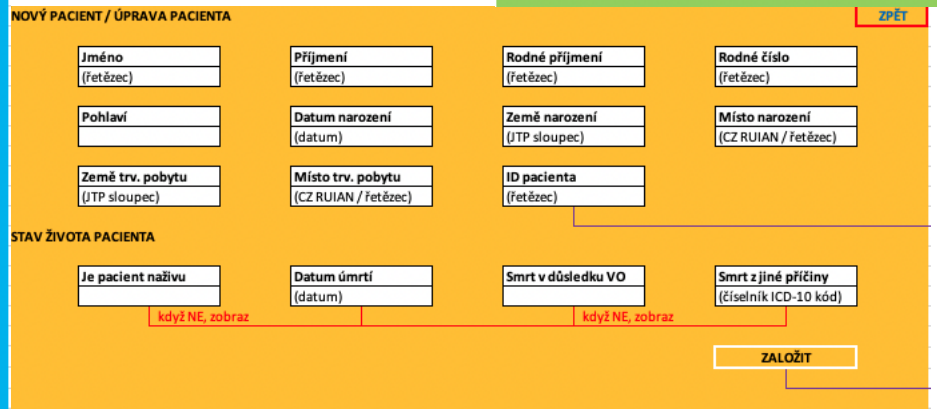
- Analytické výstupy za 1. rok
  - Návrh optimalizace úhrady vysoce specializované ambulantní péče
  - Ochota plátců diskutovat změny úhrady podmíněna znalostí předpokládaného objemu výkonů (=počtu a spektra pacientů)
- Pokračování sběru dat 2021
  - Zpřesnění informace o dg použitím Orphakódů = začlenění do NIS s funkcionalitami jako MKN
  - Orphakódy zatím začleněny pouze v NIS Medea (VFN), male k dispozici na všech typech dokladů pro pojišťovny (01, 02, 06)
  - VÝZVA všem pracovištím ERN
    - Podání žádosti o možnost zadávat Orphakód do NIS
- Seznamy Orphakódů
  - Připravovány pro všechny ERN
  - Kontakty: Klara.Hruba@vfn.cz Jana.Batt@vfn.cz

Projekt „národního registru VO“ (RARE)

- Projekt ÚZIS ve spolupráci s odborníky
  - „Optimalizace postupů při integraci prvků elektronického zdravotnictví do procesů ÚZIS a tvorba dokumentace k informačnímu systému vzácných onemocněních v ČR“,
  - Identifikace obecného datasetu zdravotních dat (CDE) ve standartní struktuře, interoperabilní s mezinárodními registry
- Základní parametr CDE = diagnóza VO pomocí Orpha-kódu







RARE / DIGOVO – další kroky

- Pilotní zadávání základního datasetu přes testovací rozhraní
  - Všichni pacienti 5 pilotních center v období 1.1.-30.6.2022
- Analýza dat
  - Doplnění podkladů pro jednání s plátci
  - Úprava datasetu
- Finalizace podrobnějšího obecného datasetu
- Návrh specifických položek dle skupin onemocnění
- Postupné začlenění datasetu do NIS poskytovatelů vysoce specializované péče

DIGOVO

Název programu či projektu	Digitalizace a optimalizace systému zdravotní péče o pacienty se vzácnými onemocněními (DIGOVO)
Realizátor:	Ministerstvo zdravotnictví ČR Všeobecná fakultní nemocnice v Praze IČO: 00064165 DIČ: CZ00064165 U Nemocnice 499/2, 128 08 Praha 2 www.vfn.cz
Komponenta:	1.2 Digitální systémy veřejné správy
Reforma/investice:	eHealth

- Aktivita 1: Vybudování koordinačního zázemí a jednotné virtuální platformy pro VO
- Aktivita 2: Standardizace datových struktur VO, propojení s českými (národními a institucionálními) a s evropskými registry VO a se systémem interoperabilní zdravotnické dokumentace v rámci eHealth
- Aktivita 3: Integrace standartního datasetu VO do nemocničního informačního systému
- Aktivita 4: Vytvoření metodiky pro testování systému a následná analýza jeho funkcionalit
- Aktivita 5: Pilotní testování systému vybranými centry vysoce specializované péče o pacienty s VO
- Aktivita 6: Analýza vyhodnocení pilotního testování

NAPVO 2022-2024 – hlavní úkoly

- **Analýza systému organizace péče** o pacienty s vzácným onemocněním (VO) včetně existujících standardů péče v evropských zemích a mezinárodních doporučení.
- **Analýza současného stavu péče** o pacienty s definovanými skupinami VO v ČR a existujících národních standardů a doporučení, identifikace nedostatků.
- **Vytvoření systému mapujícího celostátní a regionální potřebu nových odborníků** na jednotlivé skupiny VO na pracovištích národní sítě zapojených v systému komplexní sdílené péče o pacienty s VO
- **Vytvoření návrhu komplexní sdílené péče** včetně přechodu mezi dětskou a dospělou specializovanou péčí (přechodová péče)
- **Integrace zdravotní a sociální péče** o pacienty s VO
- **Vytvoření návrhu hodnocení kvality** vysoce specializované a specializované péče o pacienty s VO

Postup práce a financování

- Výzva OPZ 6/2022, realizace 2023-2025, cca 20 mil Kč
- Přípravné práce – k zahájení dnes
- Pracovní skupiny pro jednotlivé skupiny VO (ERNy)
  - Koordinátor skupiny: Zástupce ERN v MeKoVO
  - Členové: 1 zástupce každého centra, v případě 2 pracovišť v centru 2 zástupci (dětská a dospělá část), zástupce odborné společnosti (specialista mimo ERN), zástupce pacientů, PLDD / PL
- „Globální pracovní skupina“
  - Koordinátoři skupin + zástupce MZ + zástupci plátců
    - Formulace obecných návrhů společných pro všechny skupiny VO
    - Formulace návrhu legislativních opatření