European Reference Networks

ERNs – SUCCESS STORIES

Co-funded by the EU

Cancer & Family Care for Ukraine Childhood Cancer Patients - PaedCan

As of September 10, 2024, the European Paediatric Oncology Community (SIOPE, CCI-E and many ERN PaedCan Sites) assisted 1643 Ukrainian pediatric cancer patients through the SAFER (Supporting Action for Emergency Response) Ukraine program (St. Jude).

- The following countries have accepted the following number of patients under their care since 2022:

❖ Poland > 400 patients	❖ Spain > 80 patients	❖ Austria, Belgium, Romania, Slovakia: 10 - 20 patients each
❖ Germany > 250 patients	❖ Czech Republic > 70 patients	❖ Bulgaria, Croatia, Denmark, Lithuania, Portugal, Sweden: 1- 6 patients each
❖ Italy > 200 patients	❖ Switzerland and France > 60 patients	
❖ Netherlands > 100 patients	❖ United Kingdom: over 20 patients	

- 724 patients were directly evacuated through the SAFER Ukraine referral pathway.
  The other patients received support e.g. through translation of medical records.
- This data does not include patients and families who left Ukraine seeking care on their own/through other pathways. The total number of patients who left Ukraine is estimated > 2000 childhood cancer patients.

After the attack on July 8th, 2024 to the Okhmatdyt National Children´s Hospital in Kyiv, Ukraine, acute evacuation of paediatric cancer patients was needed again. ERN PaedCan members closely collaborated with SAFER Ukraine a/o directly with European National Health Ministries supporting the safe evacuation of 13 patients:

❖ Germany = 7 patients	❖ Switzerland = 2 patients	❖ Austria = 4 patients

UKRAINE – ERN-EYE

Creation of the ERNs website erncare4ua.eu and a logo

European Reference Networks

#ERNcare4Ua

Rare Diseases Doctors

Short paper published in The Lancet Regional Health

Contact with Ukrainian clinicians to identify their needs

Launch of an ERN workgroup

UKRAINE - EPICARE

European Reference Networks

#ERNcare4Ua
Rare Diseases Doctors

EPICARE WITH UKRAINE WAR

EpiCARE is officially represented at the “Emergency and Crisis Response Task Force” for Ukraine, created by the International League Against Epilepsy.

Monthly calls and direct contact with epilepsy experts and patient advocates in Ukraine.

A dedicated webinar was organized by EpiCARE with invited speakers from Ukraine.

Managing Epilepsies in Crisis Situations – The Ukraine Experience

March 21, 2024 5:00 pm

Andriy Dubenko, Volodymyr Kharytonov

See the video

EMERGENCY TASK FORCE ACTIVITIES https://www.ilae.org/files/dmfile/emergency-and-crisis-task-forces-annual-report-2023.pdf

During 2023 – 2024 the Task Force met at least once per quarter and completed the following projects:

- Drafted a suggested process for identifying crisis or emergency situations that may require a response
- Assisted in the transfer of emergency or crisis response information from the Ukraine portion of the ILAE and EpiCARE websites to the crisis response section
- Reviewed material on the crisis response webpage for completeness or correction
- Developed and presented a seminar on emergency response during the 35th International Epilepsy Congress held in Dublin, Ireland in 2023 and the European Epilepsy Congress held in Rome in 2024.

COVID-19: Helpline for Rare Bone Diseases

COVID-19 Helpline (24/7)
for supporting patients with rare bone diseases and centres during COVID-19 emergency

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

Irish Journal of Medical Science (1971 -) (2021) 190:1243–1244
https://doi.org/10.1007/s11845-020-02400-6

LETTER TO THE EDITOR

The line between COVID-19 pandemic and rare bone diseases

Luca Sangiorgi[1,2] · Evelise Brizola[1] · on behalf of the COVID-19 Helpline for Rare Bone Diseases Group

Received: 23 September 2020 / Accepted: 15 October 2020 / Published online: 2 November 2020

The recent outbreak of COVID-19 pandemic had a dramatic global impact for healthcare systems and required a rapid re-arrangement of the priorities. In Europe, a disease is defined as rare when it affects no more than 5 in 10,000 people [1]. Between 6000 to 8000 distinct rare diseases exist today affecting around 6–8% of the population—over 30 million people in Europe are directly involved [2], which number is close to the number of people currently affected with COVID-19 global-

Helpline for Rare Bone Diseases" [5]. The purpose is to provide experience and knowledge about RBD to patients and healthcare professionals working in the intensive care units and/or COVID-19-devoted wards who are treating or will treat patients affected by RBD, initially focusing on patients with osteogenesis imperfecta. For all patients with RBD, it is crucial to know that they can constantly rely on their primary care physicians and keep these professionals informed about their

Sangiorgi et al., 2021

POSITION STATEMENT Open Access

Providing high-quality care remotely to patients with rare bone diseases during COVID-19 pandemic

E. Brizola[1], G. Adami[2], G. I. Baroncelli[3], M. F. Bedeschi[4], P. Berardi[5], S. Boero[6], M. L. Brandi[7], L. Casareto[1], E. Castagnola[8], P. Fraschini[9], D. Gatti[2], S. Giannini[10], M. V. Gonfiantini[11], V. Landoni[12], A. Magrelli[13], G. Mantovani[14,15], M. B. Michelis[6], L. A. Nasto[6], L. Panzeri[5], E. Pianigiani[1], A. Scopinaro[16], L. Trespidi[17], A. Vianello[18], G. Zampino[19] and L. Sangiorgi[20*]

Abstract

During the COVID-19 outbreak, the European Reference Network on Rare Bone Diseases (ERN BOND) coordination team and Italian rare bone diseases healthcare professionals created the "COVID-19 Helpline for Rare Bone Diseases" in an attempt to provide high-quality information and expertise on rare bone diseases remotely to patients and healthcare professionals. The present position statement describes the key characteristics of the Helpline initiative, along with the main aspects and topics that recurrently emerged as central for rare bone diseases patients and professionals. The main topics highlighted are general recommendations, pulmonary complications, drug treatment, trauma, pregnancy, children and elderly people, and patient associations role. The successful experience of the "COVID-19 Helpline for Rare Bone Diseases" launched in Italy could serve as a primer of gold-standard remote care for rare bone diseases for the other European countries and globally. Furthermore, similar COVID-19 helplines could be considered and applied for other rare diseases in order to implement remote patients' care.

Keywords: 2019-nCoV, Bone diseases, Care, Coronavirus, COVID-19, ERN, Rare diseases, Remote

Brizola et al, 2020

IMPACT ON PATIENT CARE – SUCCESS STORY

COVID-19

Received: 29 May 2020 | Accepted: 29 July 2020
DOI: 10.1111/ctr.14063

BRIEF COMMUNICATION

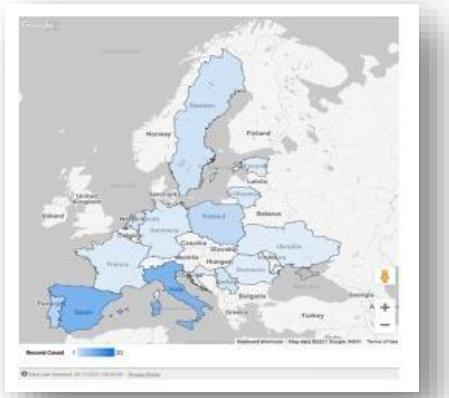
Clinical TRANSPLANTATION WILEY

Pediatric transplantation in Europe during the COVID-19 pandemic: Early impact on activity and healthcare

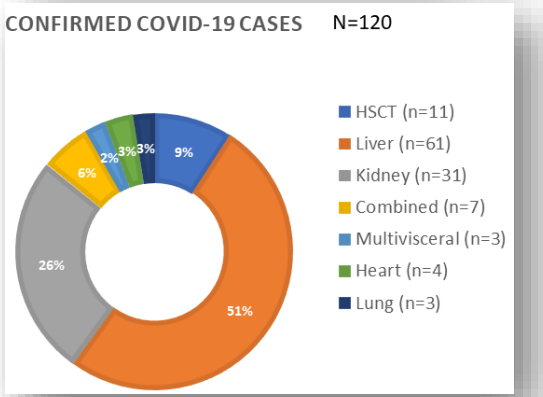
https://doi.org/10.1111/ctr.14063

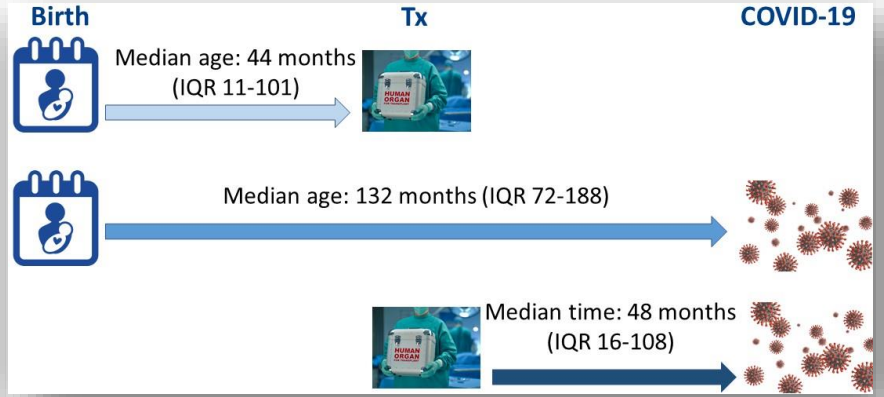
Luz Yadira Bravo Gallego[1,2,3], Mara Cananzi[4], Daniele Donà[5], Elisa Benetti[6], Marta González Vicent[7], Esteban Frauca Remacha[8,9], Paloma Jara Vega[1,8,9], *on behalf of COVID-19 study group of the Clinical Audits Working Group - ERN-TransplantChild.*

[1]ERN-TransplantChild, La Paz University Hospital, Madrid, Spain; [2]Lymphocyte Pathophysiology in Immunodeficiencies Group, Hospital La Paz Institute for Health Research (IdiPAZ), Madrid, Spain; [3]CIBERER U767, Center for Biomedical Network Research on Rare Diseases, Madrid, Spain; [4]Unit of Paediatric Gastroenterology, Digestive Endoscopy, Hepatology and Liver Transplantation, Women's and Children's Health, University Hospital of Padua, Padua, Italy; [5]Pediatric Infectious Diseases Department, Women's and Children's Health, University Hospital of Padua, Padua, Italy; [6]Pediatric Nephrology, Dialysis and Transplant Unit Department, Women's and Children's Health, University Hospital of Padua, Padua, Italy; [7]Hematopoietic Stem Cell Transplantation Unit, Department of Pediatrics, Hospital Infantil Universitario Niño Jesús, Madrid, Spain; [8]Pediatric Hepatology Department, La Paz University Hospital, Madrid, Spain; [9]Molecular Hepatology Group, Hospital La Paz Institute for Health Research (IdiPAZ), Madrid, Spain.



- 93% (112/120) were living in an EU-MS:
  - Spain: 27.5%
  - Italy: 26.7%
  - Portugal: 11.7%
- 61.7% (74/120) were male.





Conclusions

- This study reports one of the largest series of COVID-19 in pediatric transplant recipients.
  - The importance of being a member of scientific associations or networks, like the ERN-TransplantChild.
- While this population is theoretically more at risk for severe illness from SARS-CoV-2 infection due to ongoing immunosuppression and/or compromised immune system, our data show that, in this patient group, COVID-19 is mainly asymptomatic or mild, and seldom associated with patient death or graft loss.

Drug access /availability – A Joint Effort to Ensure Access to treatments for patient – ERN BOND

During the **ERN BOND Italia meeting**, the healthcare professionals highlighted critical shortages and unavailability of essential drugs and diagnostic agents

Once the needs were identified from each Italian HCP, ERN BOND engaged **AIFA** (Italian Medicines Agency) to discuss potential solutions and guarantee access to essential treatments and diagnostic agents for patients with rare diseases.

Following the meeting with AIFA, two supply solutions were identified

Importation: Via the USMAF (Maritime, Air, and Border Health Office), following the rules outlined in the Italian Ministry Degree of 11/02/1997.

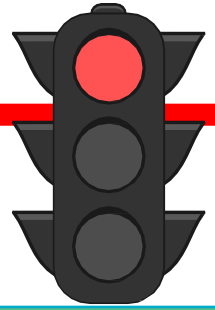
Galenic production: Sifap (Italian Society of Compounding Pharmacists) can produce these medications.

ENSURING DRUG SUSTAINABILITY - METABERN

European Reference Network | MetabERN
European Reference Network for Hereditary Metabolic Disorders

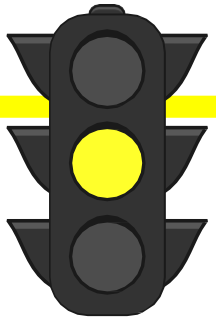
A LIFE SAVING DRUG IS WITHDRAWN

- Cobalamin C defect (CBLC) is a rare congenital disease affecting the metabolism of vitamin B12 (cobalamin), it is lethal if not treated
- Life-saving therapy: daily administration of high dosage of hydroxocobalamin (OHB12)
- In 2022 the marketing of OHB12 was discontinued and a shortage of the drug was recorded
- Patients without treatment are exposed to serious clinical events including death



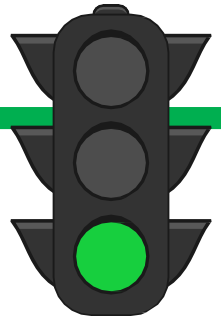
METABERN ITALY TAKES ACTION

- The Italian CBLC APS Patient Organization and the METABERN ITALY TEAM lead by the Bambino Gesù Children's Hospital in Rome join efforts among patient organisations, clinicians and the Italian Medicines Agency (AIFA).
- MetabERN Italy spreads the issue at EU level and a survey is to look for possible alternative therapeutic options.
- Results: the only therapeutic option is a OHB12 10 mg/2 ml which need to be imported in a very limited amount from Spain.



PATIENTS ARE SAFE

- Given the shortage of the drug, the Military Pharmaceutical Chemical Institute of Florence, Italy, committed by law to find solutions in national emergency needs, has taken action to make available new stocks of OHB12, for all families needing treatment.
- The shortage is restored, patients ARE SAFE



INTER-ERN COOPERATION – ERN BOND

HRpQCT

Feasibility study the potential applications of HRpQCT (High Resolution peripheral Quantitative Computed Tomography) in bone disorders related to endocrine conditions

Education and Training Survey: A Collaborative Effort enhancing education in Rare Bone diseases

Mapping of education and trainings needs and gaps in rare bone and mineral diseases in ERN BOND and EndoERN to eventually develop a comprehensive cross-border educational programme in rare bone and mineral diseases.

Preliminary phase development of molecular diagnostic tests in collaboration with MetabERN

Diagnostic characterisation of Ollier's disease

Joint Meeting EndoERN, ERKNet, ERN BOND.
"Phosphate Imbalance Disorders: Dysregulation of the FGF23A Endocrine System. Holistic management of the disease"
The Italian members of these three ERNs together with experts in X-linked hypophosphatemia (XLH) met with the aim of improving access to quality healthcare for XLH patients.

Joint Meeting EndoERN, ERKNet, ERN BOND

INTERERN COOPERATION – EURO-NMD

- The interERN Gene therapy webinar series

  The Gene therapy webinar series, led by EURO-NMD and co-organized with ERN-RND and EpiCARE, featured 14 expert speakers from 8 countries* and included 12 sessions focused on sharing lessons learned and practical implications relevant to all diseases of interest to the participating ERNs.

  *Spain, UK, Germany, USA, Netherlands, Canada, Italy, France

- The interERN Survey on Gene therapy practices in Europe

  This survey aimed to assess the current landscape of gene therapies in Europe, focusing on access conditions, organizational aspects, and clinical decision-making processes. The findings are intended to improve and harmonize practices, potentially leading to initiatives such as the establishment of treatment eligibility boards for gene therapies, with a focus on diverse disease groups.

  It was disseminated to the 5 participating ERNs (EURO-NMD, ERN-RND, EpiCARE, ERN-EYE, MetabERN) as well as centers in Ukraine, UK and Switzerland.

  185 responses were collected from 27 countries (61 EURO-NMD, 42 RND, 31 EYE, 25 EpiCARE, 25 MetabERN).

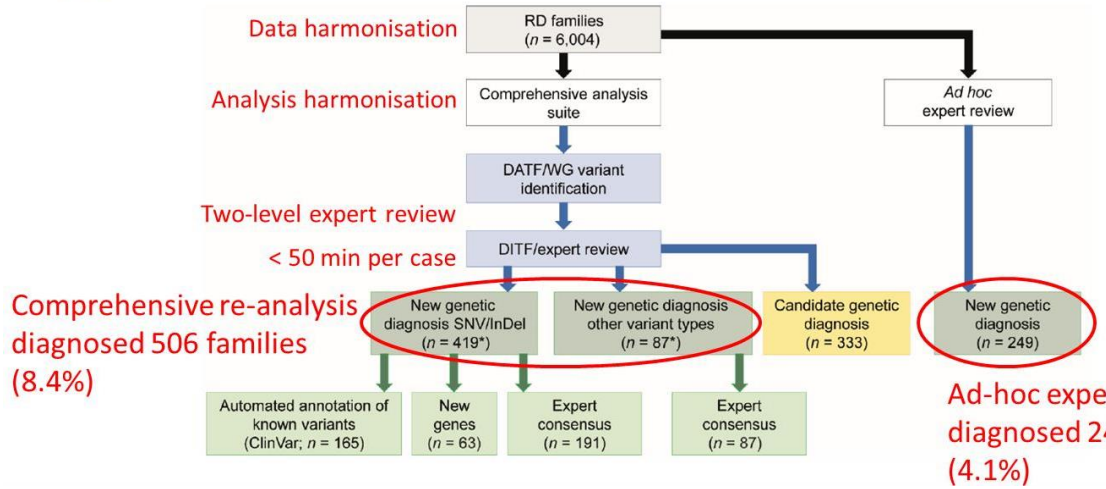
- The interERN Guideline on safe medication in mitochondrial epilepsy

  5 ERNs; 24 European experts addressed a cross-disease challenge and provided a guideline to solve a treatment dilemma (PMID: 38576261)

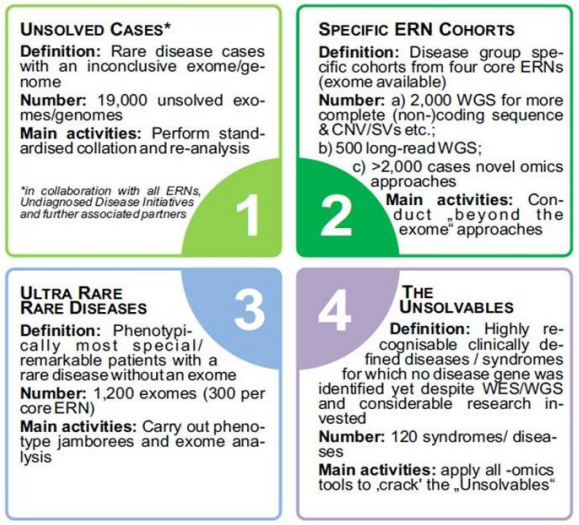
SOLVE-RD. ERN-BASED DIAGNOSTIC COLLABORATIVE RESEARCH (WWW.SOLVE-RD.EU)

- **Solve-RD network contains six ERNs: RND, NMD, ITHACA, Genturis, EpiCare, Rita**
- **Proven value of ERN-based systematic cutting-edge diagnostic research**
- **Analysis framework combined with a two-level expert review is a practical blueprint for re-analysis efforts on a global scale**

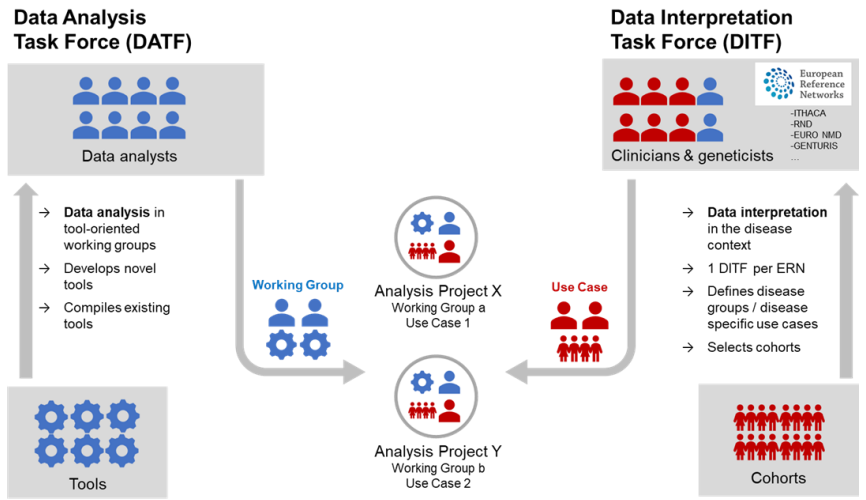
Re-analysis: yield

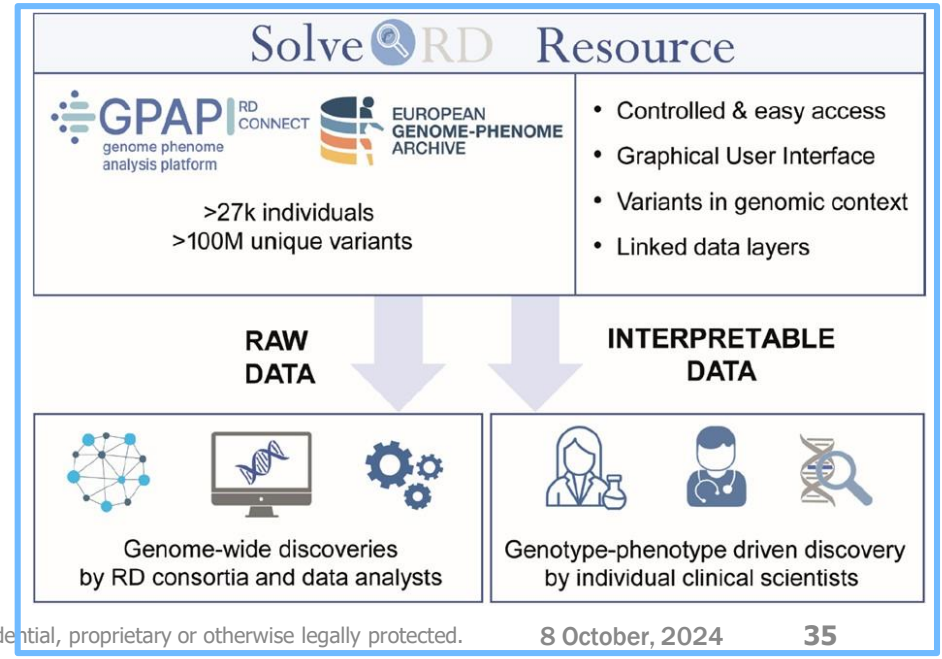


Solve-RD cohorts



Solve-RD data analysis organisation





MULTIDISCPLINARY TEAM - EPICARE

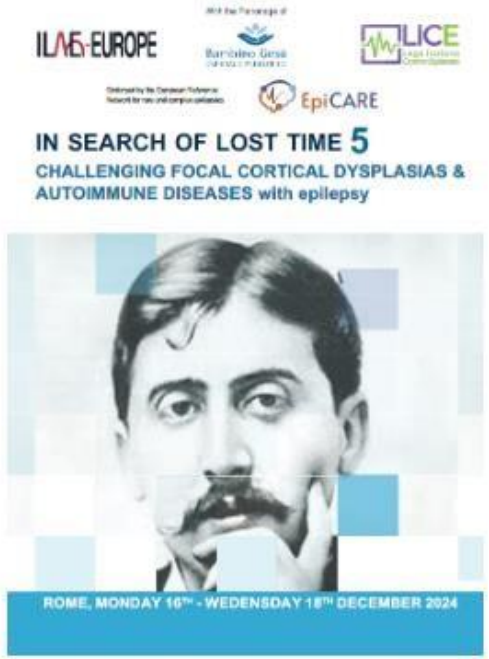
EU NETWORK OF NURSES, EEG TECHNICIANS & PARAMEDICS

Nurses, EEG technicians and neuropsychologists play an essential role in epilepsy care. For this reason, EpiCARE **reserved the first Exchange Program funded by the EU to those disciplines**. A workshop on exchange of local practices was held in 2023 leading to the creation of an EU network of NURSES and EEG Technicians involved in epilepsy care.

IN SEARCH OF LOST TIME

We are at the 5th edition of a multidisciplinary annual 3 days workshop “In search of Lost time” focusing on ne knowledge in the field.

Organized by the Italian EpiCARE members it is held in Rome and systematically involves next generation experts, researches, senior clinicians and patient advocates.

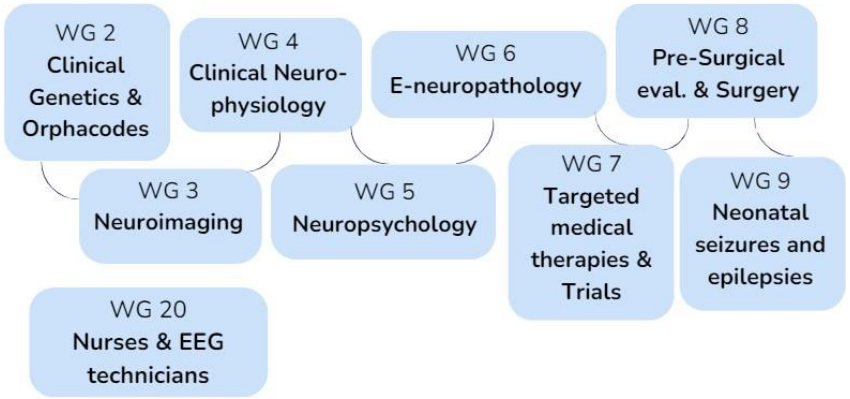


EpiCARE STRUCTURE

The need for a multidisciplinary care in epilepsy is reflected in the ERN EpiCARE structure.

WGs per discipline or field of expertise regularly interact to ensure a multidisciplinary approach of best practices in epilepsy care.

Work with EpiCARE for
Standards & Best practices



EpiCARE

European Reference Network
for rare or low prevalence complex diseases

THE FIRST ACCREDITED E-LEARNING PROGRAMME ON IMDs - METABERN

Diagnostic, Clinical & Therapeutic Education Programme

on Inherited Metabolic Disorders

Created by the

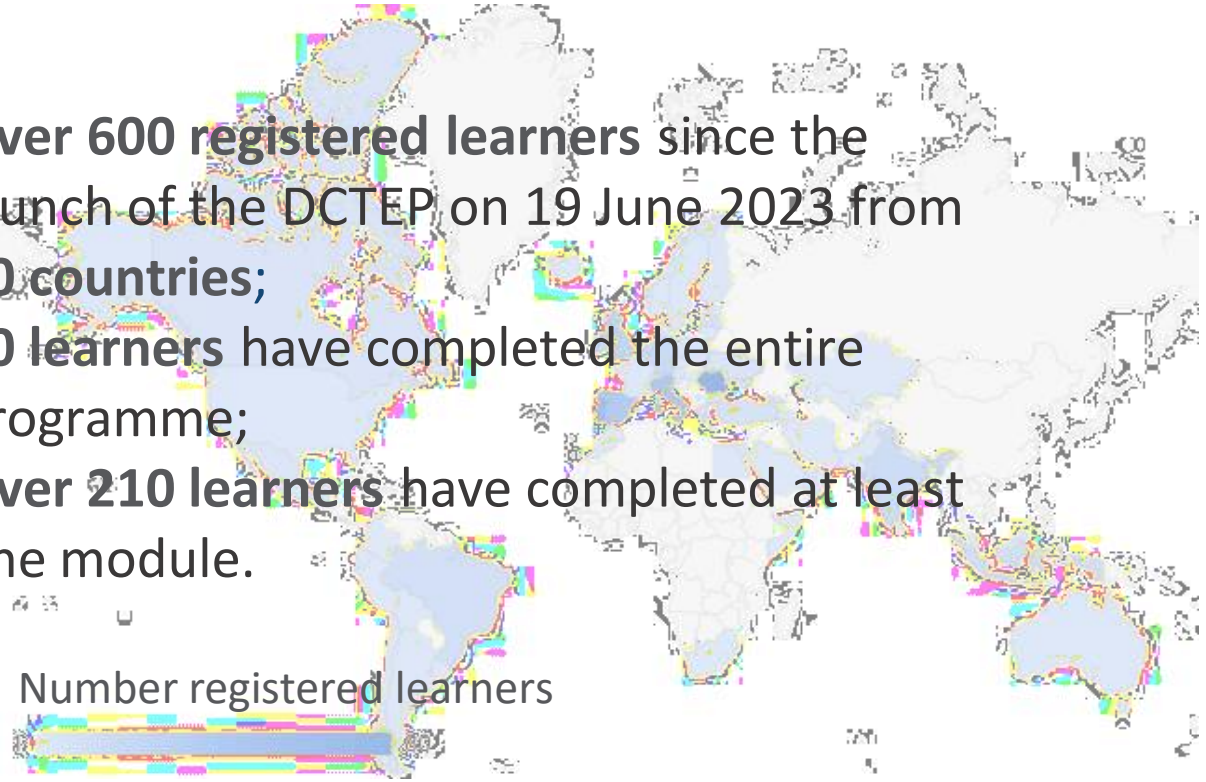
European Reference Network | MetabERN
European Reference Network for Hereditary Metabolic Disorders

Accredited by the

European Accreditation Council for Continuing Medical Education (EACCME)

11 Modules, **27** Web lectures, **17** EACCME credits.

- **Over 600 registered learners** since the launch of the DCTEP on 19 June 2023 from **70 countries;**
- **50 learners** have completed the entire programme;
- **Over 210 learners** have completed at least one module.



LEARNERS' STORIES

There were some hours really well spent, I felt like participating to an international congress, but in the quiet of my home, taking notes, rewinding and listening again

CRISTINA POPESCU
PAEDIATRICIAN

The extensive range of subjects covered by the DCTEP provide a good basic knowledge on metabolic pathways, the diseases that may affect these pathways, as well as the work-up of common presenting symptoms

MARK WIJNEN
INTERNAL MEDICINE RESIDENT

ERKUCATION: FIRST STRUCTURED ERN POSTGRADUATE CURRICULUM - ERKNET

Clinical experience

2 years
in the field of
rare kidney diseases

Webinars

3 years
every 2 weeks

including
Webinar-related
exams

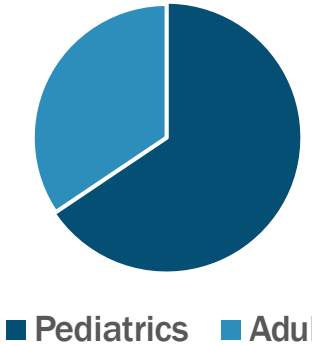
54 topics
pediatric &
adult diseases

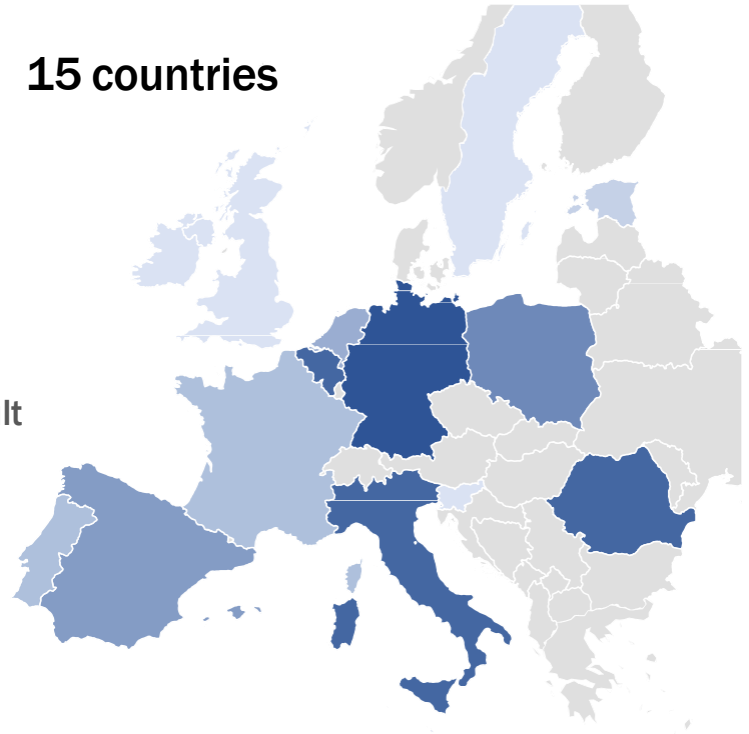
Requirements:
Attendance to 80% of ERKnet Webinars
≥ 75% correct answers in the exams

eLearning cases

topic related
cases
basic &
complex tests

Requirements:
Processing of 80% of all cases
≥ 75% correct answers





4 Classes: 347 students from 65 ERKNet HCPs in 22 countries
| 104 webinars | 31 eLearning cases | 8 workshops / CME courses

First class graduated in 12/2023:
63 „European Rare Kidney Disease Specialists“

CPMS based Cross-Border healthcare pathways in ERN-RND

(323 case discussions since 2017)

- **Neurorehabilitation**

confirming shared guidelines or discussion where no experience is known for best decision making

- **MLD treatment eligibility panels**

High-risk & high-cost: gene therapy/stem cell transplantation

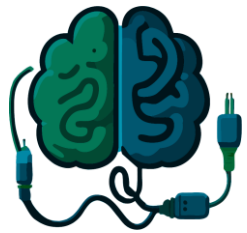
Collaboration with MLDi, (registry of treatment outcome/natural history); Lead: Amsterdam, NL

the MLD initiative

- **DBS for Dystonia**

Homogenization of a highly specialised, invasive treatment.

Lead: Würzburg, DE



- **Disease Group specific**

Consultations & best practice: Diagnosis, Disease management.

Cases clustered by the 6 ERN-RND disease groups

unsolved/complex cases

- **Neuroradiology advice**

Standard pathway for receiving second opinion on imaging findings by neuroradiology experts

Lead: Lübeck, DE

Development of future pathways
e.g. treatment eligibility for further gene therapys

EURACAN SUCCESS STORY - VIRTUAL MULTIDISCIPLINARY TUMOUR GROUPS (MDTs)

EURACAN provides financial support to MDTs to review complex or very rare patient cases registered on the Clinical Patient Management platform (CPMS).

By bringing together leading experts in different countries the goal is:

- to discuss cases of rare adult solid cancers
- to consider all perspectives and give timely and accurate diagnoses
- to increase access to novel treatments and clinical trials.

As of September 2024, the Network reviewed 286 rare adult patient cases

THE EXAMPLE OF THE RARE GYNECOLOGICAL CANCER GROUP

260 patients cases reviewed since 2017

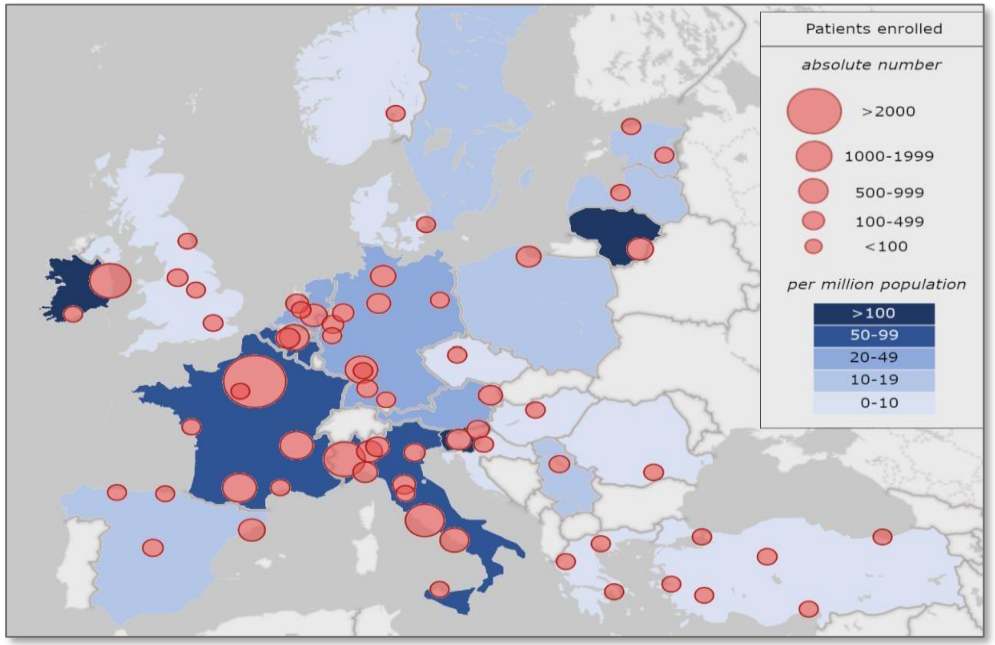
- Impact of these Tumour Boards on patient care[1]:
  - Number of reviewed patients doubled over 6 years
  - Further diagnostic testing in 1/3 of patients
  - New treatment opportunities to those originally planned for 50% of patients
  - Adherence to these treatment recommendations 94%.
  - Surveillance instead of adjuvant chemotherapy was recommended in 17% of patients
  - 37 patients gained access to off-label therapies, 4 were enrolled in clinical trials abroad

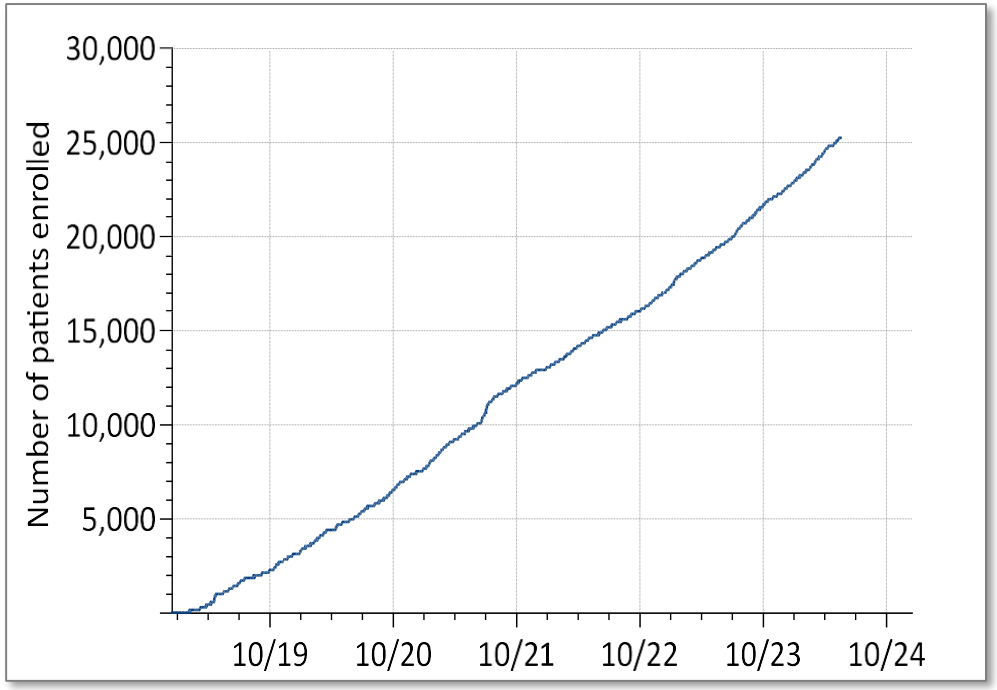
Patients could access off-label therapies not yet approved for rare gynaecological cancers, which would otherwise not have been accessible in some countries.

[1] *Alice Bergamini, ESMO Gynaecological Cancers Congress 2024 #ESMOGynae24, Abstract 82MO*

ERKReg The European Rare Kidney Disease Registry

- Centralized online registry
- **Modular concept**: Core registry and disease-specific subregistries
- >26,00,500 patients enrolled since 1/2019 in **109 specialized units in 25 countries**
- 60% pediatric and 40% adult patients
- **100 new patients added per week**
- Annual follow-up achieved in 75%
- **Key performance and outcome monitoring system**





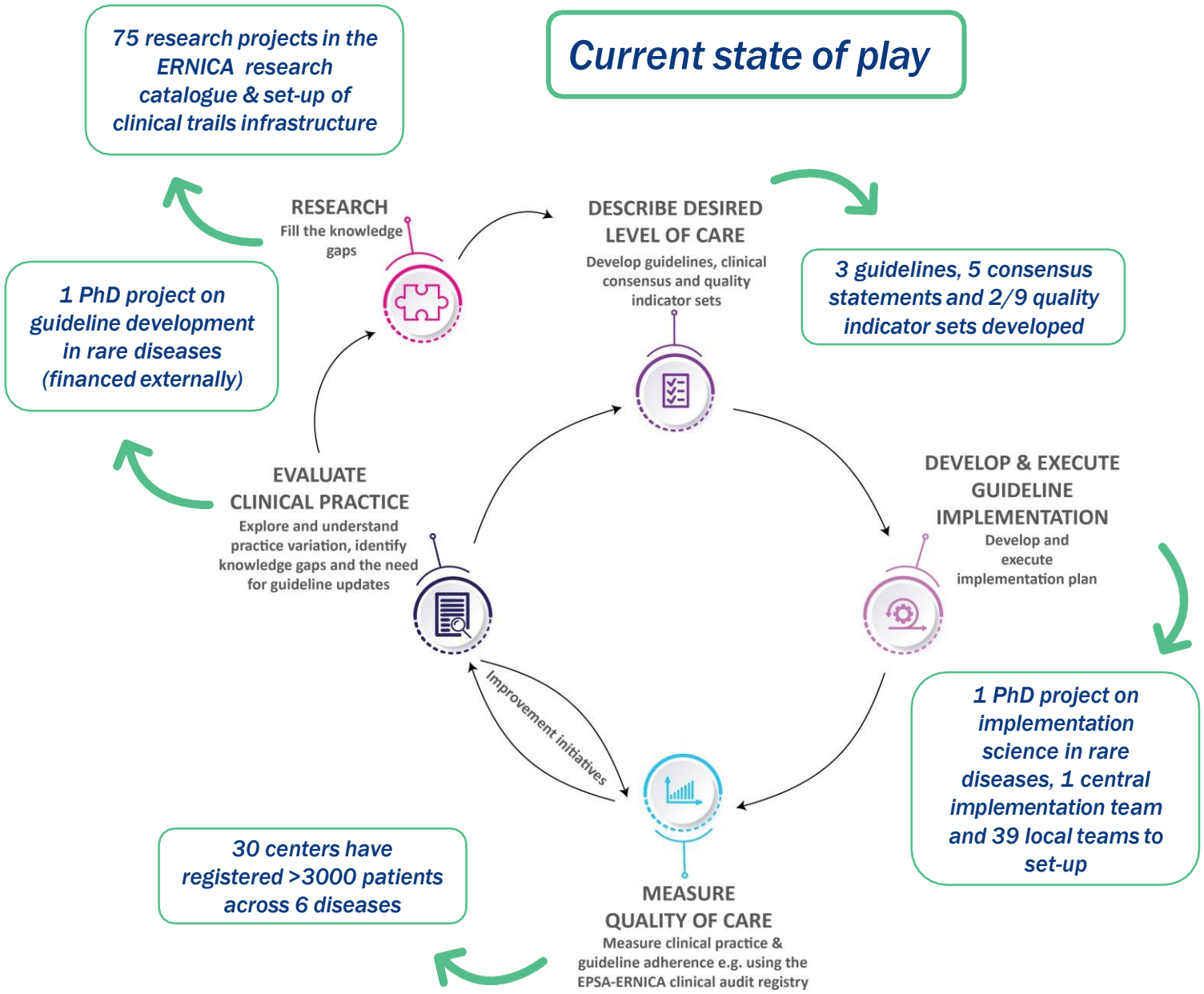
www.registry.erknet.org

Bassanese et al. Orphanet J Rare Dis (2021)
https://doi.org/10.1186/s13023-021-01872-8

ERNICA Quality Cycle

- Aim of European Reference Networks = to share, care & cure

- ERNICA has developed its own **quality cycle** to meet these aims, involving guideline development, implementation, evaluation in the patient registry and fill knowledge gaps in research ➔ ongoing process



GUIDELINES/CARE PATHWAYS - EPICARE

A DEDICATED WG ON NATIONAL HEALTHCARE PATHWAYS IN EPILEPSY CARE

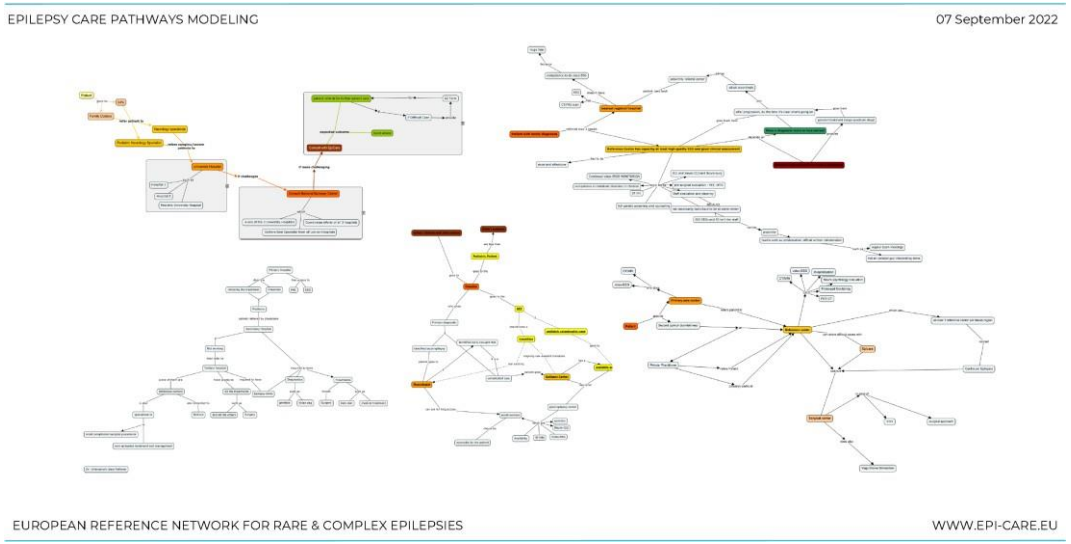
Initiated in 2021 the WG focus on public healthcare issues. Structured interviews of epilepsy leaders demonstrated that epilepsy care pathways differ significantly across EU countries, indicating the urgent need for a more structured approach.

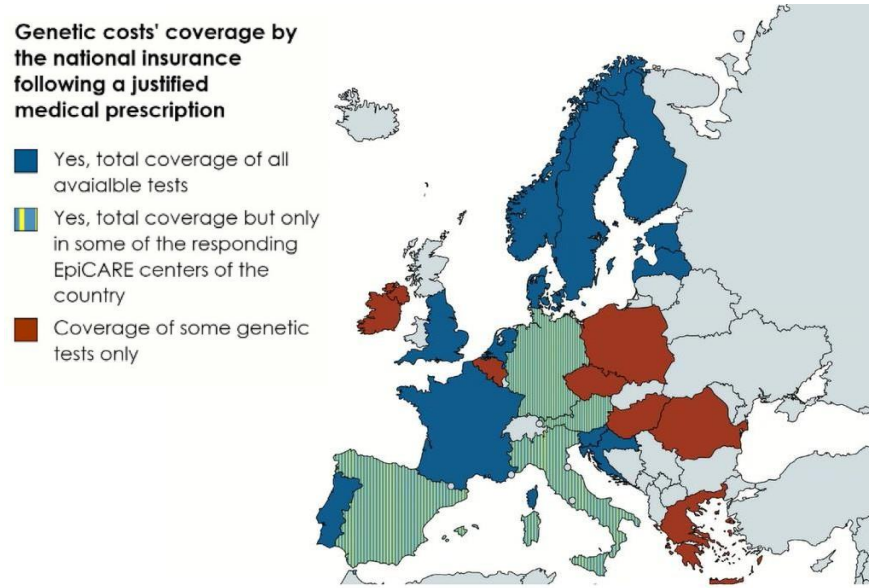
The WG is now preparing a Delphi study to reach consensus on optimal healthcare pathways for patients with epilepsy and an evidence-based definition of Level II and Lever IV reference center characteristics

INEQUALITIES IN ACCESS TO CARE:

EpiCARE performed and published a study on accessibility, availability and costs within the EU, related to genetic testing for rare epilepsies (DOI: 10.1002/epi4.12930). Differences are significant indicating the urgent need for concrete and concerted actions.

The ERN EpiCARE initiated and signed **a Memorandum of Understanding with all epilepsy-related scientific societies** (International League Against Epilepsy; European Academy of Neurology; European Paediatric Neurology Society) for **a shared production of Guidelines and Recommendations**.





"Illustration of differences in healthcare pathways in EU countries"

GUIDELINES AND CARE PATHWAYS – ERN LUNG

- 3 level approach to cross-border care even for Undiagnosed patients (Working group w/ JARDIN)
- Level 1- EXABO Online expert advice
- Level 2 – CPMS Panel discussion
- Level 3 – Cross-border referral

BREATHeREGISTRY

Population registry – Patient oriented, voluntary entering of patient details by patients.

July 2024 – 160 newly entered patients

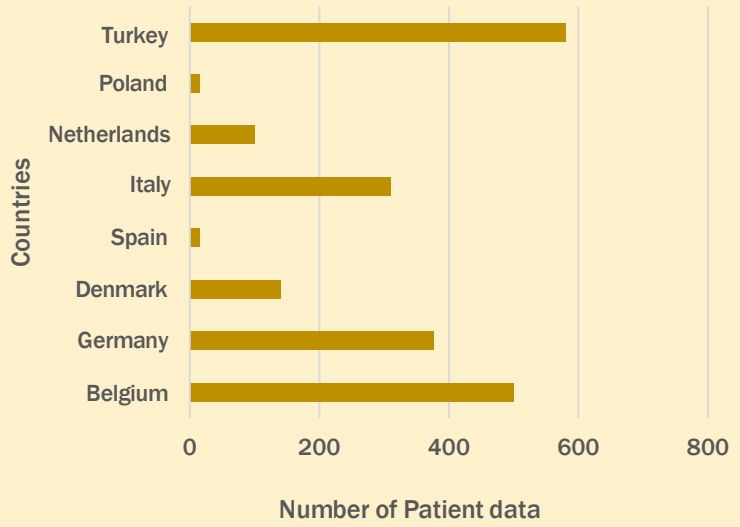
Guidelines and Patient Pathways

- 6 guidelines endorsed by ERN LUNG
- 109 publications by ERN-LUNG members
- Patient journeys for 4 ERN-LUNG disease areas
- Patient Priorities project for 3 core networks (SARC, ILD, BE)

- Patient Pathways available for CN:
  - Idiopathic Pulmonary fibrosis
  - Pulmonary Hypertension
  - Sarcoidosis
  - Primary Ciliary Dyskinesia

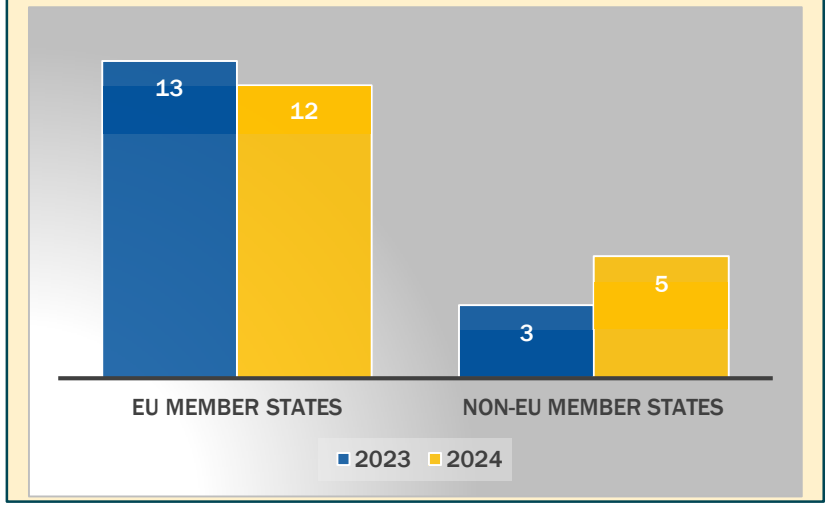
ERN-LUNG PRIME Registry

- Approx. 2000 patients in registry



ERN-LUNG Academy

- 2023 – 43 participants
- 2024 – 41 participants
- 2025 – Recruiting ongoing



Clinical Trial Network

9 research projects supported, and 3 core networks supported CF, AATD, PCD) implemented CTN. Others like BE, PH are in early phase.

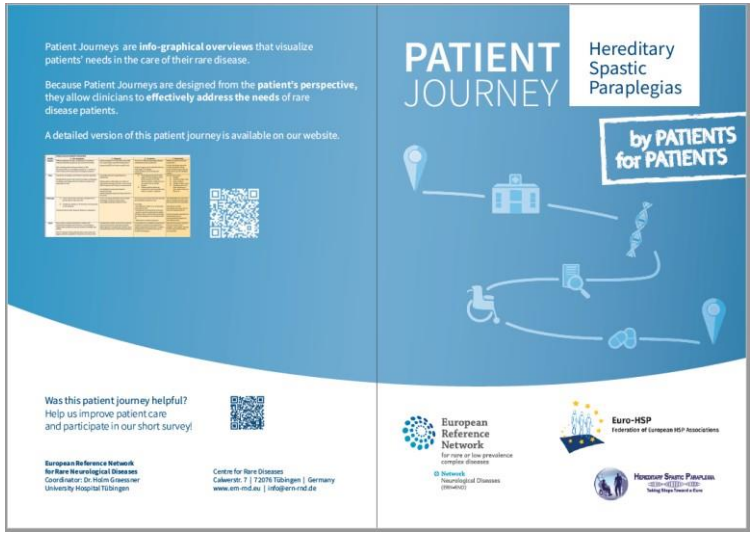
Go East Initiative

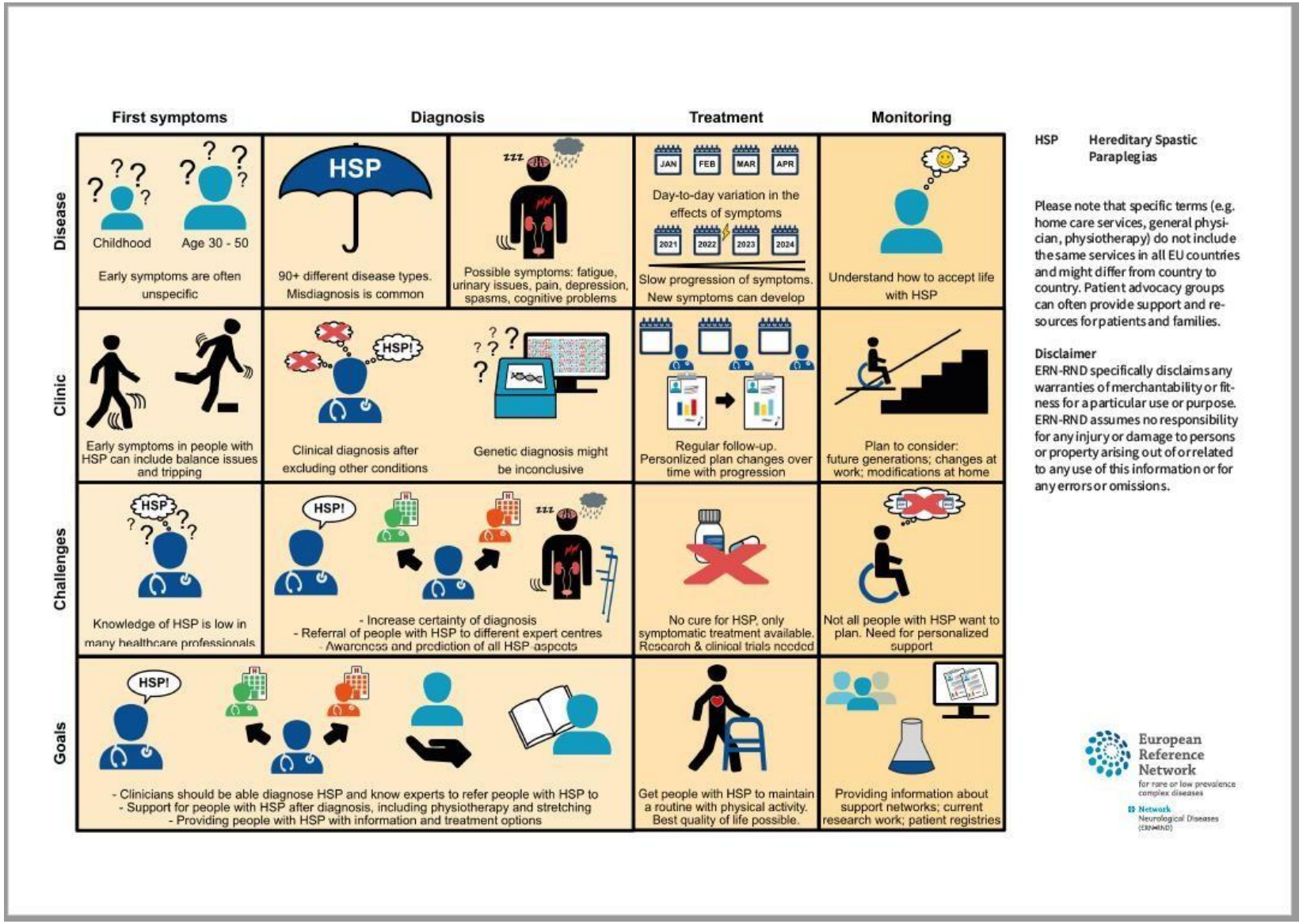
Initiative to involve under-represented Eastern European countries in ERN-LUNG. Interested HCPs located in Romania, Hungary, Slovakia.

European Reference Network
for rare or low prevalence complex diseases
Network Respiratory Diseases (ERN-LUNG)

PATIENT JOURNEYS – ERN-RND

- 5 patient journeys available and 3 in development
- Main European languages
- Goal: hand out a suitable patient journey to all newly diagnosed patients





PATIENT EMPOWERMENT - RARE-LIVER YOUTH PANEL

"We focus on improving medical care for young patients and strive to make their journeys easier by supporting, connecting and accepting."

10-15 young people with rare liver diseases (aged 18-30 years)

Meeting at least 6 times per year online and once in person

Closely involved in activities of ERN RARE-LIVER and the transition working group, participate in workshops, online meetings and development of guidelines

Decisions are made together, within the Youth Panel

- To represent the **interests of young people** and to create awareness especially in young professionals.
- To be **role models** and mentors for younger patients.
- To **connect** young people with rare diseases.

European Reference Network
for rare or low prevalence complex diseases

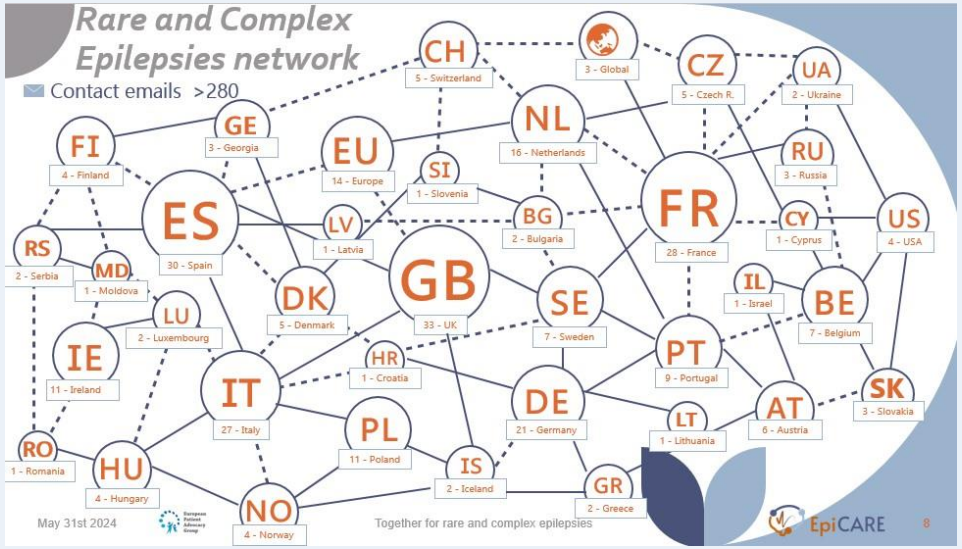
ERN RARE-LIVER

PATIENT EMPOWERMENT - EPICARE

EPILEPSIE(S) PATIENT ADVOCATES

Because of the major differences, in care and prognosis, between the different types of the epilepsies, patient representation has always been highly complex and challenging.

Despite those difficulties the EpiCARE patient advocates group succeeded in initiating partnership links with several associations across Europe.



PATIENT JOURNEY AND LEAFLETS TRANSLATIONS

EpiCARE experts together with patient representatives already produced a significant number of aetiology-specific leaflets, with information for caregivers but also for family doctors. They also take care to translate them into as many languages as possible to reach a widest audience.

Rare epilepsies Leaflets

Patient and caregivers leaflets are developed to give precise and accessible informations on rare and complex epilepsies. With **one part for healthcare profesionnals, and one part for patients and their families or carers**, these documents detail comprehensively what to expect when facing a rare epilepsy, and how to manage care.

Unless mentioned otherwise, all leaflets are in english. We are working on translating them in as many languages as possible with the help of patients associations all over Europe, so check back regularly!

Here you can download Leaflets in different languages:

Patients & caregiver leaflets

European Patient Advocacy Group

- **Dravet Syndrome**

Versions: English (EN) / Italiana (IT) /Română (RO) / Hrvatska (HR) / Deutsche (DE) / Norsk (NO) / Srpska (RS) / Svensk (SE)

- **Hypothalamic Hamartoma**

Versions: English (EN) / Roman (RO) / Hrvatska (HR)

- **Ring Chromosome 20**

Versions: English / Hrvatska (HR) / ελληνική έκδοση (GR) / Lietuviška (LT)

- **Alternating Hemiplegia of Childhood**

Versions: English (EN) / Hrvatska (HR) / Italiana (IT) / Española (ES) / Française (FR)

- **GLUT1 Deficiency Syndrome**

Versions: English (EN) / Hrvatska (HR) / Română (RO) / Deutsche (DE) / Italiana (IT)

- **CDKL5 Deficiency Disorder**

Versions: English (EN) / Española (ES) / Portuguesa (PT) / Hrvatska (HR) / Română (RO)

- **Lennox-Gastaut Syndrome**

Versions: English (EN) / Română (RO) / Hrvatska (HR)

- **SYNGAP1**

Versions: English (EN) / Română (RO) / Hrvatska (HR)

- **RETT Syndrome**

PATIENT JOURNEYS & LEAFLETS

PATIENT EMPOWERMENT: OPEN DIALOGUE BETWEEN PATIENTS AND CLINICIANS

Workshop: "Patient priorities in ERN BOND beyond Quality of Life Provision of care to RBDs patients"

4 topics explored in the rare bone disease area:

- Pain management
- Pregnancy
- Movement/Functional limitations
- Transition from pediatric to adult

Commentary paper on the workshop results under submission

Publication "Defining priorities in the transition from paediatric to adult healthcare for rare bone disease patients: a dialogic approach" in the ERN BOND special issue in EJMG (Scognamiglio et al., 2024)

First author, a rare bone disease patient, awarded as "*Italian Health Champion*" aiming to valorize important scientific achievements in the biomedical field

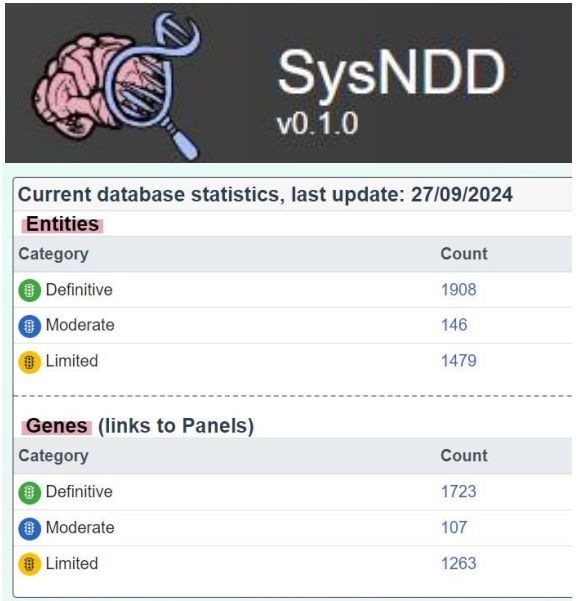
RESEARCH - ITHACA

- Elevating Care and Research in Genetic NeuroDevelopmental Diseases (NDD), a Great Success
  - Second-of-its-kind European workshop focused on the complex care and research of genetic neurodevelopmental disorders.
  - Over 250 experts including clinicians, patients, and researchers from across Europe
  - Held at the University Institute of Lisbon (ISCTE), April 4-5, 2024
  - Highlights of interdisciplinary collaboration and patient-centric approaches that led to practical innovations in diagnostics and therapies
- EuroNDD 2024 aligned with and supported by ERN ITHACA's ongoing initiatives to enhance patient care across Europe

SUPPORT TO ORPHANET - ITHACA

- Orphanet is represented in ITHACA's board

- Orphanet summaries
  - Each HCP is invited to collaborate with ORPHANET to create or update entries of the European catalogue for (neuro)developmental disorders
  - Orphanet proposes yearly a list of missing/outdated summaries that are delivered on a voluntary base by ITHACA members
  - Over 80 contributions online since 2020
- Link between Orphanet and the SysNDD database
  - SysNDD is a database of genes involved in Intellectual disability and Autism
  - ITHACA support SysNDD, its curation and funded the link between Orphanet and SysNDD for the list of non-syndromic ID genes (over 2000)
- Orphanet nomenclature & HPO ontology
  - ITHACA has contributed to several updates in the ontology used by ORPHANET, based on Human Phenotypic Ontology database
  - Main contributions are in the field of fetal pathology and recent update in the classification of spinal dysraphisms



ORPHANET AND HPO REVISION – ERN-EYE

- **Meetings**

→ 3 meetings already took place: December 2023 & February 2024 in Strasbourg & July in Amsterdam

- **Revision and follow-up on ontologies**

→ since 2018 (ERN-EYE meeting in Mont Ste-Odile, France)

Sergouniotis et al. Orphanet Journal of Rare Diseases (2019) 14:8
https://doi.org/10.1186/s13023-018-0980-6

Orphanet Journal of Rare Diseases

LETTER TO THE EDITOR — Open Access

An ontological foundation for ocular phenotypes and rare eye diseases

Panagiotis I. Sergouniotis[1*], Emmanuel Maxime[2], Dorothée Leroux[3], Annie Olry[2], Rachel Thompson[4], Ana Rath[2], Peter N. Robinson[5], Hélène Dollfus[3,6*] and for the ERN-EYE Ontology Study Group

- **Context**

→ Development of a registry for Rare Eye Diseases (REDgistry)

- **Context**

→ Improvement of genetic diagnosis

OUTCOMES WILL BE INTEGRATED IN THE CURRENT ORPHANET CLASSIFICATION & HUMAN PHENOTYPE ONTOLOGY (HPO)

human phenotype ontology

orphanet

Develop Child and Orphan Device Evaluation support for rare diseases

European Reference Network | ERN eUROGEN Rare Urogenital Diseases & Complex Conditions

DeCODe

Aim

1. Support development of paediatric and orphan devices for rare diseases
2. Consortium co-funded by the EC, of academics, developers, clinicians, regulatory experts, funding experts, in-silico testing experts
3. ERN eUROGEN and MetabERN involved

Goal

1. Develop a critical path for orphan device development
2. Provide support to 5 orphan device developers
3. Call for proposals in March 2025

Consortium

1. Academics, developers, clinicians, regulatory experts, funding experts, in-silico testing experts
2. Co funded by EC
3. ERN eUROGEN and MetabERN involved

DeCODe development

1. Concept development
2. Advanced development
3. Device development and prototyping
4. Testing and certification
5. Implementation/Lifecycle management

DeCODe support

1. Business (IP, Needs assessment, Value proposition, Funding)
2. Technology (Proof of concept, Infra testing, Hardware & software, Connectivity)
3. Regulatory (Preparation, Quality assessment)

Pilot cases

1. 5 cases
2. Rare and paediatric
3. Unmet need, potential for significant benefit
4. Off label going towards on label use, de novo development
5. Call open in spring 2025
6. Device developers get min 6 months of support