ERN-RND Scientific Symposium

European Healthcare for RND Patients

28th October 2025
Tutzing, Germany

European Reference Network
for rare or low prevalence complex diseases

Network Neurological Diseases (ERN-RND)

www.ern-rnd.eu I info@ern-rnd.eu

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PROGRAMME

Tuesday 28 October 2025

SESSION 1

Cross-Border Access to Highly Specialized Therapies for Ultra-Rare Conditions

Chaired by Ines Hernando

TIME (CET)	TOPIC	SPEAKER
08:20	**Fetal** Surgery as an ERN example	Jan Deprest (ERN ERNICA, Leuven)
08:40	"Just like home" Program at Fondazione Telethon for **accessing Gene Therapy**	Stefano Zancan (Fondazione Telethon Milan)
09:00	EURORDIS' **Lighthouse Concept**	Ines Hernando (Eurordis)
09:20	**Krabbe Disease** Expert Panel	Pascal Martin (Tübingen)
09:40	**Panel Discussion**	
10:10	*Coffee break*	

SESSION 2

Multidisciplinary Case Discussion for Controlling Access and Ensuring Best Possible Outcome for Highly Specialized Therapies

Chaired by Ludger Schöls

TIME (CET)	TOPIC	SPEAKER
10:40	**Epilepsy** Surgery as an EpiCARE use case	Alexis Arzimanoglou (ERN EpiCARE, Barcelona)
11:00	**Deep brain stimulation** – Pitch and Live Case Discussion	Martin Reich (Würzburg)
11:30	**Metachromatic Leukodystrophy** Treatment Eligibility – Pitch and Live Case Discussion	Nicole Wolf (Amsterdam)

SESSION 3

JARDIN - Integration of ERNs into National Healthcara Systems

Chaired by Holm Graessner

TIME (CET)	TOPIC	SPEAKER
12:00	Overview and **First Results** of JARDIN	Till Voigtländer (Vienna)
12:20	**National Reference Networks**	Holm Graessner (Tübingen)
12:40	Development and implementation of **ERN care pathways**	Birutė Tumienė (Vilnius)
13:00	**National support of ERN members**	Pavla Doležalová (Prague)
13:20	**Panel discussion**	

JAN DEPREST

Professor of Obstetrics & Gynaecology at KU Leuven (Belgium) and University College London (UK)

Fetal Surgery as an ERN Example

Prenatal screening ultrasound is now universally offered across Europe and often leads to the prenatal diagnosis of congenital malformations. When such abnormalities are detected, patients are referred to tertiary centers for counseling and to determine the optimal timing and mode of delivery. Fetal surgery is rarely indicated and is only offered to fetuses who are at risk of death or irreversible organ damage without prenatal intervention. Currently, there are two conditions for which level I evidence demonstrates that fetal surgery improves outcomes. However, because these conditions are rare and the learning curve for most procedures requires 30–60 cases, training and maintaining a skilled multidisciplinary team is challenging. Consequently, these highly specialized surgeries may not be available locally, and patients may need to travel internationally for treatment.

Europe offers a unique situation in which, in most cases, patients can undergo surgery abroad if it is not available locally—provided the necessary administrative and insurance paperwork is completed. However, the interval between diagnosis and intervention is typically short, and the anxiety induced by the prenatal diagnosis of a serious congenital malformation often creates a de facto emergency situation.

Using two examples, severe congenital diaphragmatic hernia and spina bifida aperta, to illustrate the treatment, we describe how our center in Belgium has developed an efficient and patient-centered workflow, which includes:

1. Rapid, personalized communication between our team, the referring prenatal diagnosis center, and the family.
2. Secure, patient-driven creation and transfer of medical records and data.
3. Routine teleconferencing for the initial consultation, allowing for personalized counseling prior to first travel.
4. Consolidation of all required prenatal investigations into a single day prior to intervention is possible.
5. Minimal hospital admission, postoperative outpatient surveillance, and direct access to the fetal surgery team in case of complications.
6. Ongoing, direct communication with the referring center to ensure continuity of care.

The presentation will also cover the limitations and challenges of this procedure.

Jan Deprest MD PhD is Professor of Obstetrics & Gynaecology at KU Leuven and UCL. He has a research interest in fetal surgery, in particular in antenatal interventions that modulate fetal development in case of severe congenital birth defects.

He is founder of Eurofoetus, bringing together fetal medicine specialists and manufacturers to design fetoscopic instruments now widely used. The consortium completed two randomized trials in fetal surgery, one on the treatment of twin-to-twin transfusion syndrome and on diaphragmatic hernia. Awarded fellowship ad eundem at the RCOG (2012), the International Society of Ultrasound in Obstetrics and Gynecology Ian Donald Award (2018), international membership in the US National Academy of Medicine (2022), the Eric Saling Award from the World Association of Perinatal Medicine (2023), International Society for Prenatal Diagnosis Pioneer in Prenatal Diagnosis and Therapy Award (2024).

STEFANO ZANCAN

Head of Clinical Development and Operations
in Fondazione Telethon (Italy)

“Just Like Home” - Program at *Fondazione Telethon* for Accessing Gene Therapy

Presentation and description of Fondazione Telethon ‘s program Just Like Home, a program to support families and patients to overcome all obstacles and come to specialized clinical centre in Italy to receive gene therapy. Discussion on the meaning of “access to therapy” and considerations on its concrete applicability.

Stefano Zancan has been working in clinical research for more than 20 years, mainly in GSK but also in Novartis and Aptuit, managing a portfolio of clinical projects and leading a group of scientists accountable for the end to end operational management of the assigned clinical studies from concept protocol to final reporting. He is experienced in data protection in clinical trials, quality and GCP.

He is currently the head of Clinical Development and Operations in Fondazione Telethon, contributing to move projects from basic research to clinical research and leading subsequent clinical operations activities.

He is also the Head of the Just Like Home Program, a program Fondazione Telethon has set to support patients get access to Gene Therapies.

INES HERNANDO

Healthcare Director, EURORDIS (Spain)

EURORDIS' **Lighthouse Concept**

The presentation will discuss the opportunity to establish an EU model to commission certain highly specialised services through closer EU cooperation to ensure the delivery of timely, safe, accessible, and sustainable highly speciliased cross-border services. Taking as a starting point the concept of access to healthcare services, the presentation will discuss the European Lighthouses model leading to the centralisation of certain highly specialised healthcare services at EU level and outline the policy pathway that policymakers could use to assess the costs and benefits of such a reform.

Inés Hernando joined EURORDIS as ERN and Healthcare Director in February 2018. In this role she is responsible for monitoring the European landscape on rare disease healthcare from a policy and advocacy perspective, managing all related activities.

Previously she worked for two years at COCIR as eHealth Senior Manager where she led the organisation's work on all topics related to digital health, focusing specifically on issues such as data protection, interoperability and cross-border data flows. She also worked for eight years in the Spanish Electronic Health Record initiative as eHealth Coordinator where she provided overall programme management and help build the programme partners' ecosystem that included all Regional Health Authorities and the Ministry of Health.

PASCAL MARTIN

Senior Physician at the Department of Neurology and Epileptology at the University Hospital of Tuebingen (Germany)

KRABBE Disease Expert Panel

Krabbe disease is an ultra-rare, often devastating lysosomal storage disorder for which hematopoietic stem cell transplantation (HSCT) remains the only disease-modifying treatment option. However, the decision to proceed with HSCT is complex, particularly in late-onset or atypical cases, due to heterogeneous clinical courses and limited outcome data.

To address these challenges, the ERN-RND has launched a Krabbe Treatment Eligibility Panel (TEP) — a structured, CPMS-based expert forum designed to harmonize treatment decisions and collect evidence across Europe. The TEP enables cross-border case discussions, integrates patient data into the Tübingen Krabbe Registry, and fosters consensus-building on treatment criteria and monitoring strategies.

Complementary initiatives include a European survey to map current diagnostic and therapeutic practices, and the systematic prospective inclusion of discussed patients for longitudinal evaluation. Together, these efforts aim to standardize care, reduce insecurity in treatment decisions, and advance clinical research readiness for future therapeutic developments in Krabbe disease.

Dr. Pascal Martin has been serving as a Senior Physician at the Department of Neurology and Epileptology at the University Hospital of Tuebingen since March 2019. He began his clinical career in September 2012 as a resident physician. His scientific focus is on MR imaging in leukodystrophies in Prof. Samuel Gröschel's working group at the children's hospital at the University of Tübingen.

Together with Prof. Samuel Gröschel, Lucia Laugwitz, and Tamara Martin, he coordinates the Krabbe Treatment Eligibility Panel, which was launched in March 2025.

ALEXIS ARMAZINOGLOU

ERN EpiCARE Coordinator, Director Epilepsy Program,
Hospital Sant Joan de Déu, Universitat de Barcelona (Spain)

Epilepsy Surgery as an epiCARE use case

Improved knowledge of the natural history of the epilepsies, the introduction of new techniques for the assessment of their causes and origins, such as the early identification of localized brain lesions, have progressively led to an earlier and more frequent use of surgical therapy for patients with focal onset epilepsies. Lately, although a number of neurologists unfortunately still consider epilepsy surgery a “last resort” treatment, considerable progress has been made for a better definition of eligibility criteria and the most appropriate investigations to be performed.

We will demonstrate the complexity of surgical case discussions, briefly highlighting the following points:

- Competences in both neurology and clinical epileptology, practicing within a multidisciplinary team, represent the foundations of a comprehensive epilepsy surgery program.
- When considering epilepsy surgery in children, the neurodevelopmental issues are at least as important as achievement of seizure freedom.
- Considering a patient as a candidate to surgical treatment is not the starting point but the result of a long road of presurgical evaluation.
- Early referral to specialized centres offers the patients a chance for an optimal diagnostic workup.
- Misconceptions of physicians and the public are still a major barrier, preventing timely referral to epilepsy centres for surgical assessment.
- A good knowledge of the natural evolution of each aetiology (type of lesion) at the origin of seizures and the corresponding risks from a specific surgical approach are prerequisites for a shared and timely decision-making process.
- It is the combination of results of a panel of investigations, appropriately chosen and properly interpreted by a multidisciplinary team of experts, that can lead to improvement of seizure control and quality of life of children operated on because of their epilepsy.

Professor Alexis Arzimanoglou is, since 2019, the elected Coordinator of the European ReferenceNetwork for Rare and Complex Epilepsies, ERN EpiCARE. Since October 2023, he is also the Director of the Epilepsy Program at the Barcelona Children’s Hospital San Juan de Dios, Universitat deBarcelona, Spain.

In 1999, together with an international group of colleagues, he launched the first epilepsy journal, *Epileptic Disorders,* that included video material, since 2014 recognized as the official educational journal of the International League Against Epilepsy (ILAE).

For his global contribution to epilepsy care he received in 2007 the “ILAE-Epilepsy Ambassador award” and in 2022 the “ILAE-Europe Education award”.

MARTIN M. REICH

Professor for Molecular Neuroimaging
at the University Hospital Würzburg (Germany)

Deep Brain Stimulation -
Pitch and Live Case Discussion

Martin Reich will introduce the audience to the ERN-RND online multidisciplinary board for Deep Brain Stimulation (DBS) in Dystonia. On a real-life example he will highlight which questions may arise before and after treating a patient with this invasive surgical procedure, and how the multidisciplinary expert board can help making the best decisions for a patient. Finally, he will give an outlook on how to harmonize DBS procedures for dystonia patients in Europe.

Prof. Martin M. Reich is a neurologist and international expert in deep brain stimulation (DBS) and molecular neuroimaging. He leads the visualDBS Lab and serves as Senior Attending Neurologist at the University of Würzburg. A former researcher fellow at Harvard Medical School, he also holds a guest professorship at EPFL Geneva and is the ERN-RND speaker for DBS in dystonia. His award-winning research focuses on brain network mapping, DBS programming, and AI-based neurotechnologies. With over 50 publications and 2,400+ citations, Prof. Reich continues to drive innovation in personalized neuromodulation for movement disorders.

TILL VOIGTLAENDER

Associate Professor in Neurosciences
at the Medical University of Vienna (Austria)

Overview and **First Results of JARDIN**

In the European Joint Action on Integration of ERNs into National Healthcare Systems (JARDIN), we aim to enhance the impact of the ERNs even further by addressing the current gaps between them and the different healthcare systems in the EU Member States and Norway. In this context, it is of particular importance to take into account the diversity of the systems, as well as the various aspects that contribute to the functioning of the ERNs. Therefore, JARDIN is producing recommendations and tools that are adaptable to the needs of the participating countries and cover a number of important topics, such as the national governance of healthcare providers participating in an ERN, quality assurance models, patient pathways and ERN referral systems, the formation of national reference networks and undiagnosed disease programs, improvement of data management, and the identification of national support options for ERN-HCP. In addition, strategies for systematic dissemination of information about ERNs are being developed, with a specific emphasis on patients as well as the medical community. Also, it is of utmost importance to ensure the sustainability of the proposed actions and implementations, which should be integrated into updated national plans and strategies for rare diseases.

JARDIN started in January 2024 and will therefore soon go into its third year. Most work packages dedicated the first project year to the evaluation of the current status quo in the aforementioned fields in participating countries. Currently, we are in the process of analyzing existing gaps and developing concrete solutions how to close them. The main focus of the last part of the project will then be to run pilot implementations in some countries to test these solutions, and to support capacity building in all countries in order to translate recommendations into concrete actions.

Till Voigtländer, MD, Associate professor in Neurosciences at the Medical University of Vienna, board-certified consultant in neurobiology with more than 20 years of professional experience in neurobiological laboratory medicine and neuropathology. He has been Orphanet country coordinator since 2004. In 2008 he organized a petition for a RD national plan that was presented to the Austrian Minister of Health. In response to this petition, he was officially assigned head of a committee that was mandated with the elaboration of the outlines of this plan. On RD day 2009, he organized the first walk for RD in Vienna, and in 2010 the first Austrian congress for RD. In 2011 he initiated the foundation of the umbrella organization of RD patient organizations and the National Coordination Center for RD (NCCRD) in Austria. As head of the NCCRD, he led the drafting of the Austrian National Action Plan for RD, which was published in 2015. He later became head of the newly founded National Office for RD in Austria. He is past and present member of several national and international boards and expert groups related to RD healthcare policies (EUCERD 2010-2013, CEGRD 2014-2016, Cross-border Healthcare Directive Expert Group 2012-2013, ERN BoMS since 2015, advisory board on RD and advisory board on newborn screening to the Austrian MoH). He was co-chair of the ERN BoMS from 2015 to 2018 and 2021 to 2024. Currently, he is coordinating the European Joint Action on Integration of ERNs into National Healthcare Systems (JARDIN).

HOLM GRAESSNER

Managing Director of the Rare Disease Centre,
Coordinator of the European Reference Network
for Rare Neurological Diseases (Germany)

National Reference Networks

The JARDIN WP7 work package focuses on developing National Reference Networks (NRNs) for rare and complex diseases across Europe.

Tasks include analyzing existing national structures, developing recommendations, designing models for NRNs, and implementing pilot projects. A current state survey provided the information that in at least ten EU countries NRN exist.

A mixed-methods approach combining survey, focus groups, and national consensus processes was used to develop overarching recommendations for National Reference Networks in the European Union and Norway. Over 400 stakeholders contributed to the Target State Survey, informing national recommendations. Focus group meetings were organized in five countries. Consensus processes used an adapted Delphi methodology to refine country-specific NRN strategies in eight countries.

Key themes of the recommendations include goals and activities, governance, funding, legal frameworks, and integration with European Reference Networks (ERNs). Recommendations stress care coordination, patient involvement, sustainability, and alignment with national health systems. Pilot implementations that are planned in Austria, Czech Republic, Estonia, Germany, Norway, Portugal, and Greece will practically test NRN models. Ultimately, results will inform best practices for embedding NRNs into European healthcare systems.

Holm Graessner has graduated in Biomedical Engineering, Cybernetics, Electrical Engineering, German Language and Literature, Philosophy as well as Business Administration. He received his PhD “Summa cum laude” in 2004 and, then, he obtained his MBA degree in 2008.

He has been Managing Director of the Rare Disease Centre, since 2010, at the University and University Hospital Tübingen, Germany. www.zse-tuebingen.de He is Coordinator (Lead PI) of the European Reference Network for Rare Neurological Diseases (ERN-RND, www.ern-rnd.eu). Together with Olaf Riess, he was leading the H2020 Solve-RD project on “Solving the unsolved rare diseases” (www.solve-rd.eu) and also co-leads the Clinical Research Network of European Rare Disease Research Alliance that has started on 1 Sept 2024. He is a founding member of the European 1 Mutation 1 Medicine initiative and member of the Board of Directors of the N-of-1 Collaborative.

In 2020, he has been appointed fellow of the European Academy of Neurology (EAN) and since 2025 he is a Commissioner in the Lancet Commission on Rare Diseases.

In 2023, he received the EURORDIS Black Pearl Award for Leadership and the first Martha and Wilfried Ensinger Award.

BIRUTĖ TUMIENĖ

Assoc. prof., Head of Unit for Clinical Genetics and Genomics
and the Head of the Coordination Center for Rare Diseases
at Vilnius University Hospital Santaros Klinikos (Lithuania)

Development and Implementation of **ERN Care Pathways**

One of the nine Joint Action JARDIN work packages (WP) is dedicated to rare disease care pathways (WP6: National care pathways and ERN referral systems). Since the development of care pathways always takes into account two sources: (1) evidence-based data, as Clinical Practice Guidelines (CPGs), collected from experts and (2) experience-based data collected from patients, care pathways may both help to implement ERN-developed CPGs and comprehensively respond to the needs of patients and families.

Finally, development of national care pathways provides the means to adapt reference care pathways (as those developed by ERNs) to the real-life legal and organizational structures of national health systems.

In the development and implementation of rare disease care pathways, JARDIN's WP6 goals are:
1) to develop methodologies for the development of national rare disease care pathways;
2) to develop a set of pilot ERN reference care pathways;
3) to investigate legal and organizational prerequisites for the development and implementation of national care pathways;
4) to provide capacity building for Member States.

In the dedicated session of ERN-RND Annual Meeting, we will discuss the results of this work obtained to date and further proceedings.

Assoc. prof. Birutė Tumienė is a clinical geneticist by background. She is the Head of Unit for Clinical Genetics and Genomics and the Head of the Coordination Center for Rare Diseases at Vilnius University Hospital Santaros Klinikos.

She is deeply involved in many European Reference Networks (ERN)-related activities, including a position of a Chair of the ERN Board of Member States. In the Joint Action on ERN integration into national systems JARDIN (https://jardin-ern.eu/), she co-leads WP6 on rare disease care pathways and leads WP4 on ERN sustainability actions, including revision of National RD Plans and Strategies in the EU.

Currently, she is a co-leader of a Workstream for education and capacity building in the European Rare Disease Research Alliance ERDERA, a member of the IRDiRC Diagnostic Scientific Committee, a National Coordinator of Orphanet Lithuania, a member of a Panel of Experts in the WHO Collaborative Global Network for Rare Diseases.

In 2021, assoc. prof. Tumienė was awarded EURORDIS Black Pearl European Leadership Award for her input into the development of European Reference Networks.

PAVLA DOLEŽALOVÁ

Professor of Paediatrics, Head of Paediatric Rheumatology Unit, Charles University in Prague (Czech Republic)

National Support of ERN Members

Despite 9 years since their official establishment, integration of ERN clinical centres into the healthcare systems of EU member states has not been completed. Major inequities exist among countries as well as individual hospitals in many aspects related to their operational characteristics.

EU4Health funded Joint Action JARDIN WP9 has been devoted to identify the main unmet needs of ERN clinical centres and formulate recommendations on their support both at the national and hospital levels. EU Survey tool was used to gather comprehensive data on the operational, educational, financial, and strategic challenges faced by the ERN clinical centres.

A key challenge identified was the extreme variability in **staffing patterns,** with most European countries lacking defined requirements for ERN centres. Many centres reported insufficient personnel to manage daily workloads, resulting in unpaid overtime for physicians and restricted services due to shortages of nursing and allied health professionals. **Education and training opportunities** for RD care also showed significant heterogeneity.

While half of the surveyed countries offer RD-specific educational programs, participation is often hindered by conflicting work schedules and limited institutional support. Structured postgraduate training and protected time for education are critical needs that remain unmet in many centres.

Financial sustainability remains a pressing issue, with only 7 countries providing dedicated public funding for ERN activities. **Quality assurance** practices for RD care are also underdeveloped. Only 37% of countries have implemented a national quality assurance model, and less than a quarter of hospitals have RD-specific mechanisms in place. **Strategic planning** for ERN clinical centres is similarly fragmented.

Based on the survey results, WP9 has identified the main areas requiring systematic support and has started a multistakeholder consultation process on the draft recommendations addressing these issues. A concerted effort is required to unify practices, secure consistent funding, and strengthen institutional support, enabling ERNs to fulfil their mission of delivering equitable and high-quality care to RD patients across Europe.

After graduation in general medicine at the Charles University Pavla Doležalová specialised in general paediatrics. The subspecialty training at the Birmingham Children´s Hospital paediatric rheumatology unit and subsequent consultant post at Great Ormond Street Hospital for Children in London built foundations to her strong relationships with the UK and wider European paediatric rheumatology community. She participated in multiple international studies and clinical trials including the European project „SHARE“, that resulted in the publication of consensus clinical practice guidelines for the main groups of rare paediatric rheumatic diseases. In the past 20 years she has been leading the paediatric rheumatology team at the General University Hospital in Prague, which qualified as a member of ERN RITA in 2017. She also chairs the subspecialty subgroup of the Czech Society of Paediatrics and the paediatric rheumatology accreditation board. She serves as a BoMS representative and medical co-lead of several national rare disease projects. These include a project on the optimisation of the complex care for rare disease patients that mirrors many of the activities of the Joint Action JARDIN.