View the web version

ERN GENTURIS Newsletter

European Reference Network | Genetic Tumour Risk Syndromes (ERN GENTURIS)

November 2024

ERN GENTURIS news

ERN GENTURIS Annual Meeting

ERN GENTURIS
General annual meeting
28 -29 November 2024, Amsterdam
Nicoline Hoogerbrugge
European Reference Network
for rare or low prevalence complex diseases
Network
Genetic Tumour Risk Syndromes (ERN GENTURIS)
ERN GENTURIS
With every diagnosis we can help an entire family
Radboudumc

This year the Annual Meeting of ERN GENTURIS took place in Amsterdam on 28 and 29 November. Nearly 70 members and Patient Representatives participated in the meeting. The meeting was full of inspiring sessions with great speakers, including the presentations of **Dr. Sangiorgi Luca, MD, PhD** (chair of ERN Coordinators Group), and of **Prof. Alexander Hoischen** (professor of genomic technologies for immune-mediated and infectious diseases) explaining the European scale rare disease research projects Solve-RD and ERDERA.

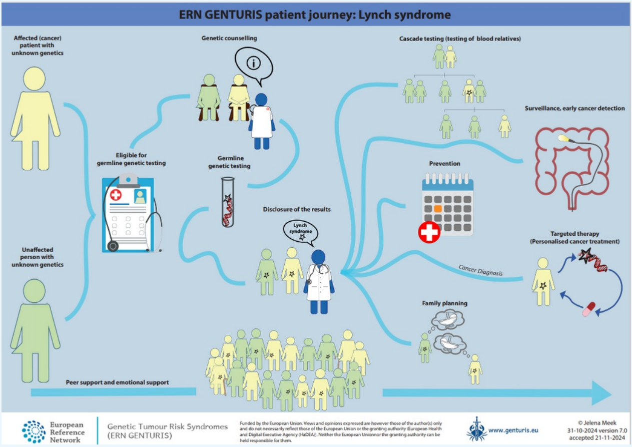
As it was a very special meeting, the Network prepared a goodbye song for **Prof. Nicoline Hoogerbrugge**, who is going to retire next year.

Interview with Prof. Nicoline Hoogerbrugge

In this interview Prof. Nicoline Hoogerbrugge shared what are her greatest achievements as ERN GENTURIS coordinator, what she learned from this experience and what she would like to pass on to others.
Read the interview:

full interview

New ERN GENTURIS Patient Journey


ERN GENTURIS patient journey: Lynch syndrome
Affected (cancer) patient with unknown genetics
Genetic counselling
Cascade testing (testing of blood relatives)
Surveillance, early cancer detection
Eligible for germline genetic testing
Germline genetic testing
Prevention
Disclosure of the results
Lynch syndrome
Targeted therapy (Personalised cancer treatment)
Unaffected person with unknown genetics
Cancer Diagnosis
Family planning
Peer support and emotional support
European Reference Network
Genetic Tumour Risk Syndromes (ERN GENTURIS)
Funded by the European Union. Views and opinions expressed are however those of the author(s) only and do not necessarily reflect those of the European Union or the granting authority (European Health and Digital Executive Agency (HaDEA)). Neither the European Unionnor the granting authority can be held responsible for them.
www.genturis.eu
© Jelena Meek
31-10-2024 version 7.0
accepted 21-11-2024

The ERN GENTURIS patient journeys are developed for **patients**. These patient journeys provide the **patient** with a first impression of what to expect. The clinical expert can use the patient journey to explain the route/journey a genturis patient will take. The details of the patient's personal journey will be filled in by the clinician (personalised care).

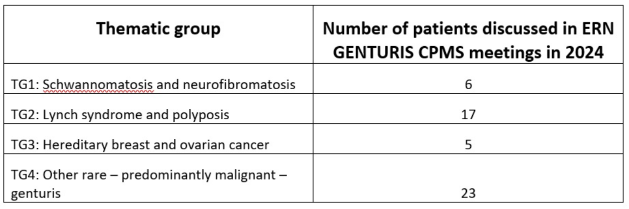
ERN GENTURIS patient journeys are infographics that consist of an accessible visual overview, supported by a detailed information matrix.

All ERN GENTURIS patient journeys include information regarding **germline genetic testing, genetic counselling, diagnosis, surveillance and follow-up, (peer) support and, where applicable, transition of care.**

ERN GENTURIS has developed 7 patient journeys so far and the Lynch syndrome patient journey was recently revised.
Find out more on our website:

patient journeys

ERN GENTURIS CPMS



Thematic group	Number of patients discussed in ERN GENTURIS CPMS meetings in 2024
TG1: Schwannomatosis and neurofibromatosis	6
TG2: Lynch syndrome and polyposis	17
TG3: Hereditary breast and ovarian cancer	5
TG4: Other rare – predominantly malignant – genturis	23

In the discussions organized through the Clinical Patient Management System (CPMS), ERN GENTURIS has provided tailored expert advice to **51** genturis patients and their families in 2024 so far.
For more information on how to refer a patient for discussion in an ERN GENTURIS CPMS meeting and how to use the CPMS, please see:

- How to refer a patient
- CPMS information and policy

CPMS 2.0 information

CPMS 2.0 transition - overview of the most important information

All ERN GENTURIS CPMS users migrated to CPMS 2.0 on **Tuesday November 26**, so their account was automatically transferred and they should be able to log in to the new CPMS immediately after the transfer.

CPMS 2.0 transition - upcoming online trainings
Currently scheduled trainings for CPMS 2.0 users, which will be updated

regularly as more trainings will become available later. Please use the links below to add the training for a specific date to your calendar.

- CPMS 2.0 web - CARE interface (intended audience - Clinicians):
    - Monday, December 2nd, 5:00 PM CET - Add to calendar
    - Thursday, December 12th, 9:00 AM CET - Add to calendar

Further details on the transition to CPMS 2.0 and all relevant documentation can be found on our CPMS 2.0 information webpage.

ERN GENTURIS Endorsed Guidelines

ERN GENTURIS endorsed guidelines for Thematic Group 2: Lynch syndrome and polyposis – polyposis syndromes

- Updated European guidelines for clinical management of familial adenomatous polyposis (FAP), MUTYH-associated polyposis (MAP), gastric adenocarcinoma, proximal polyposis of the stomach (GAPPS) and other rare adenomatous polyposis syndromes: a joint EHTG-ESCP revision, by Gloria Zaffaroni et al., British Journal of Surgery, May 2024.

ERN GENTURIS endorsed guidelines for Thematic Group 3: hereditary breast and ovarian cancer syndrome

- Risk reduction and screening of cancer in hereditary breast-ovarian cancer syndromes: ESMO Clinical Practice Guideline, by C. Sessa et al., Annals of Oncology, January 2023.

endorsed guidelines

ERN registries

24
ERN
De Europese Commissie heeft 24 ERN's aangesteld.

Registries are a very important part of the work done by ERNs. Any research on rare diseases can only take place if data is available and registries are the key as repositories of health data.

For an overview of why ERN registries are important for rare disease research (available in English, French and Dutch, including subtitles), please see newly released videos:

- English spoken version, including subtitles
- French spoken version, including subtitles
- Dutch spoken version, including subtitles

genturis registry

Meeting in Skien, Norway

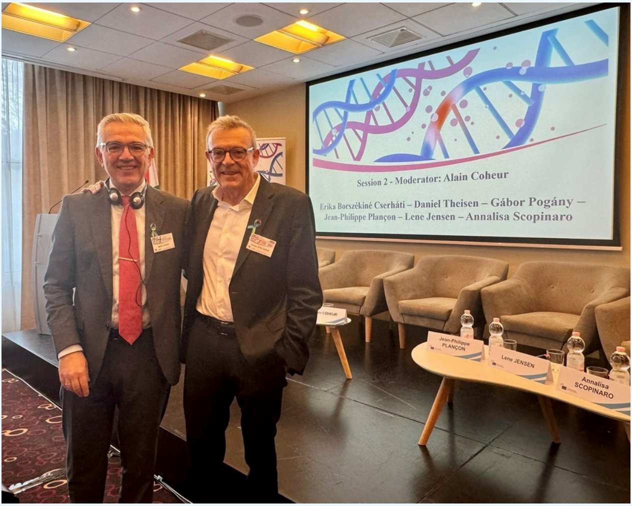
The first meeting of the **Nordic network about hereditary cancer risk** was held in Skien, Norway on 12 November 2024. It was prepared by ERN GENTURIS members, Cathrine Bjorvatn & Hildegunn Høberg Vetti, working at Haukeland University Hospital in collaboration with the Norwegian Society of Human Genetics (NSHG) and the Norwegian Society of Medical Genetics (NFMG). The meeting was inspired by and as a spin-off of ERN GENTURIS Network meetings.

"It was inspiring to see both the clinical challenges, where we had found similar solutions. But even more so, where we differed. We all go back with new inspiration on how to do better for the Nordic ERN GENTURIS patients.", said Charlotte Kvist Lautrup, working in Aarhus Universitetshospital, Denmark.

The Nordic countries have much in common in regards to both language and organisation of health care.
The next meeting is going to take place in Copenhagen, Denmark in 2025.

'For an EU commitment to tackling rare disease' - conference in

Budapest


Session 2 - Moderator: Alain Coheur
Erika Borszékiné Cserháti – Daniel Theisen – Gábor Pogány –
Jean-Philippe Plançon – Lene Jensen – Annalisa Scopinaro
Jean-Philippe PLANÇON
Lene JENSEN
Annalisa SCOPINARO

Dr. Ignacio Blanco, MD, PhD, represented **ERN GENTURIS** during the conference **'For an EU commitment to tackling rare diseases'** organised by the **European Economic and Social Committee** (EESC) together with the **Hungarian Presidency of the Council** of the EU in Budapest, Hungary on 29 November. With this event, the EESC wanted to demonstrate the commitment of civil society organisations to support the establishment of a European policy framework for rare diseases, bringing together all the stakeholders needed for a concerted strategy in the areas of research, prevention, digital, healthcare, social protection and investment in rare diseases, both at European and national levels.

More information

Education opportunities

ERN GENTURIS educational webinars

11 December 2024
Fertility preserving measures in hereditary cancer syndromes – the oncofertility perspective
Kenny A. Rodriguez-Wallberg

22 January 2025
ERN GENTURIS guidelines on constitutional mismatch repair deficiency diagnosis, genetic counselling, surveillance, quality of life, and clinical management
Chrystelle Colas & Katharina Wimmer

More on our website:

webinars

Virtual Edition of the 5th Hereditary Cancer Genetics Course 2024

ESHG
EUROPEAN SOCIETY OF HUMAN GENETICS

5th Course on Hereditary Cancer Genetics
Bertinoro, Italy | September 17-20, 2024

Organized by members of the ERN GENTURIS, this course aims at delivering up-to-date knowledge on hereditary cancers to clinical and molecular geneticists in training or certified. The online course runs from **October 1, 2024, until December 31, 2024**.

- Registration (€60 per person): LINK
- Course programme

more information

Diagnosing Rare Diseases: from the Clinic to Research and Back

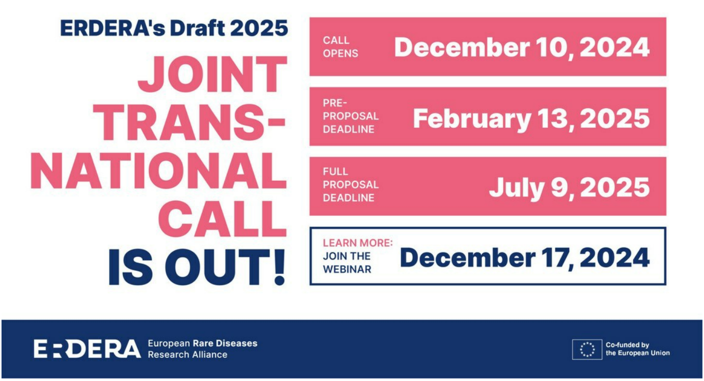
In the context of the European Joint Programme for Rare Diseases EJPRD, co-developed by Chrystelle Colas (ERN GENTURIS), Laurence Faivre (ERN ITHACA), and the Foundation For Rare Diseases, the Massive Open Online Course (MOOC) addresses the diagnostic research progress, types of genetic tests for rare diseases, and the impact of having or lacking a diagnosis on patients' lives.You will be able to discover the role of research, clinical investigation and data sharing in diagnosing rare diseases.

The MOOC is continuously open for enrollment and the access to the content is free for the first 5 weeks.

For more information check the link

course

GENERAL NEWS



ERDERA's Draft 2025

JOINT TRANS-NATIONAL CALL IS OUT!

CALL OPENS December 10, 2024

PRE-PROPOSAL DEADLINE February 13, 2025

FULL PROPOSAL DEADLINE July 9, 2025

LEARN MORE: JOIN THE WEBINAR December 17, 2024

ERDERA European Rare Diseases Research Alliance

Co-funded by the European Union

The upcoming launch of **Joint Transnational Call (JTC) for Proposals 2025** is set to officially open on **December 10, 2024,** this call will invite research teams from across Europe and beyond to submit collaborative projects focused on ***“Pre-clinical therapy studies for rare diseases using small molecules and biologicals – development and validation”.***

Find out more:

website

European Commission news

Newly released Health at a Glance:

Europe 2024 report calls for boosting healthy longevity and strengthening the health workforce

The Health at a Glance: Europe 2024 report, provides health stakeholders and policymakers with the data they need to make sound decisions about future actions and policies. The report also serves as a roadmap of progress made, as well as a compass for the direction to take in the future.

The report highlights the importance of **collaboration** – not only between health and finance ministries, but also across sectors and among countries – **to build sustainable, high-performing and people-centred health systems**. It will be essential to foster competitiveness and innovation within the health sector, positioning it as a driver of economic growth and societal resilience.

more information

Study on guidance to increase access to healthcare for people with disabilities

SURVEY
Collecting data on access to cancer care of people with disabilities
Deadline: 8 December
© European Union, 2024. All rights reserved.
Images source: iStock.com

#HaDEA #EU4Health #HealthUnion
European Commission

The European Health and Digital Executive Agency (HaDEA) has commissioned ICF S.A. to conduct a Study on guidance to increase access to healthcare for people with disabilities, as part of the EU4Health 2022 work programme.

Share your experience if you undergone any cancer screening & you are a person with disabilities in the EU, Norway, or Iceland via the confidential survey and help shape the future of accessible healthcare.
Take the survey by 8 December, available in various languages and easy-to-read formats:

survey

Funding opportunities

European Commission Calls

- HaDEA Calls for **Proposals on Health**
- HaDEA Calls for **Tenders on Health**

- Horizon Europe calls for **Funding on Health**

Upcoming Meetings & Events

ERICA GA & ERN Research Conference

The European Rare Disease Research Coordination and Support Action consortium (ERICA) 4th General Assembly & ERN Research Conference will take place on **11-13 December, 2024** in Udine, Italy.

More information

erica-rd.eu
Call for abstracts
Info & Registration
ERICA ERN Research Conference
11-13 December 2024 in Udine, Italy
Funded by the European Union
ERICA
European Reference Network
MetabERN
European Reference Networks

register here

Visit our:
Website and LinkedIn

ERN GENTURIS

With every diagnosis **we can help an entire family**

This newsletter is sent to {{email}}.
You received this e-mail because you are registered at ERN GENTURIS

unsubscribe