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ERN GENTURIS
Newsletter

Genetic Tumour Risk Syndromes
(ERN GENTURIS)

October 2024

ERN GENTURIS news

New ERN GENTURIS guideline

ERN GENTURIS GUIDELINE ON CONSTITUTIONAL MISMATCH REPAIR DEFICIENCY DIAGNOSIS, GENETIC COUNSELLING, SURVEILLANCE, QUALITY OF LIFE, AND CLINICAL MANAGEMENT

Publication date 17 October 2024

Authors: Chrystelle Colas, Léa Guerrini-Rousseau, Manon Suerink, Richard Gallon, Christian P. Kratz, Éloïse Ayuso, CMMRD Guideline Group*, Laurence Brugières, Katharina Wimmer

* Author names and affiliations are presented on the following pages

EUROPEAN REFERENCE NETWORKS
FOR RARE, LOW PREVALENCE AND COMPLEX DISEASES
Share. Care. Cure.

The ERN GENTURIS guideline on constitutional mismatch repair deficiency diagnosis, genetic counselling, surveillance, quality of life, and clinical management has been published online!

Congratulations to all authors: ***Chrystelle Colas, Léa Guerrini-Rousseau, Manon Suerink, Richard Gallon, Christian P. Kratz, Éloïse Ayuso, CMMRD Guideline Group, Laurence Brugières, Katharina Wimmer.***

To download the complete guideline, the EJHG publication, the pocket guide and the plain language summary in English, please go to the ERN GENTURIS website:

guideline

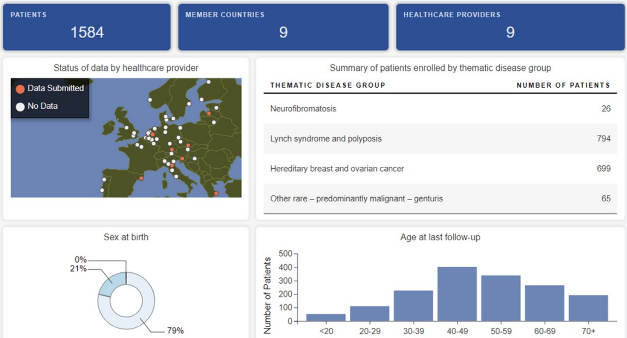
ERN GENTURIS spotlights

In this ERN GENTURIS spotlight we introduce **Prof. Kathleen Claes**, who is a clinical laboratory geneticist at Ghent University Hospital and a professor at Ghent University, as well as the PI of Cancer Research Institute Ghent. Her major interest is in molecular diagnostics for cancer predisposition syndromes.

"Within our hospital we take care of genturis patients with a multidisciplinary team. Hereby, a close link between the laboratory and the clinic is key. (...) At our biweekly meetings challenges for the workup of complex genturis cases are discussed. These also allow to streamline the multidisciplinary care and make sure our approaches remain well aligned."

read the interview

ERN GENTURIS Registry



PATIENTS 1584

MEMBER COUNTRIES 9

HEALTHCARE PROVIDERS 9

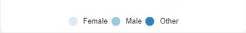
Status of data by healthcare provider

Summary of patients enrolled by thematic disease group

THEMATIC DISEASE GROUP	NUMBER OF PATIENTS
Neurofibromatosis	26
Lynch syndrome and polyposis	794
Hereditary breast and ovarian cancer	699
Other rare – predominantly malignant – genturis	65

Sex at birth

Age at last follow-up



The GENTURIS registry is a web-based platform that facilitates standardized data registration and sharing of data across Europe to improve diagnostics, treatment and disease prevention in patients with a genetic tumour risk syndrome (genturis).

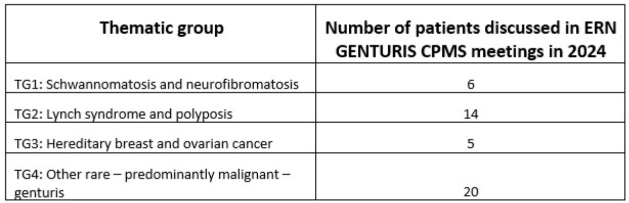
The registry enables a new wave of **research opportunities** from translational to epidemiological studies to clinical trials, and will accelerate the generation of new knowledge on genetic tumour risk syndromes.

- Since the last newsletter (September 2024) more than **336** new patients have been included in the GENTURIS registry.

For more information, click the link below or contact the UMCG team via **genturis-registry@umcg.nl**

more information

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	14
TG3: Hereditary breast and ovarian cancer	5
TG4: Other rare – predominantly malignant – genturis	20

In the discussions organized through the Clinical Patient Management System (CPMS), ERN GENTURIS has provided tailored expert advice to **45** 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 guides

CPMS 2.0 information

!IMPORTANT!
If you would like to submit patients for discussion in one of our recurring CPMS meetings after December 1st, please contact the ERN GENTURIS helpdesk manager (Jurriaan.holzenspies@radboudumc.nl) before entering the patient into the CPMS

- ***Please note: To facilitate the data migration, please close all currently open panels that have previously been discussed in an ERN GENTURIS recurring CPMS meeting*** (step-by-step guide)
- An overview of frequently asked questions regarding CPMS 2.0 is available here.

CPMS 2.0 transition - user data migration test

ERN GENTURIS is one of the first ERNs to migrate using the two-step approach (user data will be migrated first and patient data will be migrated later) and we have volunteered to help test the user data migration from CPMS to CPMS 2.0.

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):
    - Thursday, 31st of October, 9:00am CET - add to calendar
    - Monday, 4th of November, 5:00pm CET - add to calendar
- Training for CPMS 2.0 web – CARE interface (intended audience - Guests):
    - Tuesday, 12th of November, 4:00pm 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.

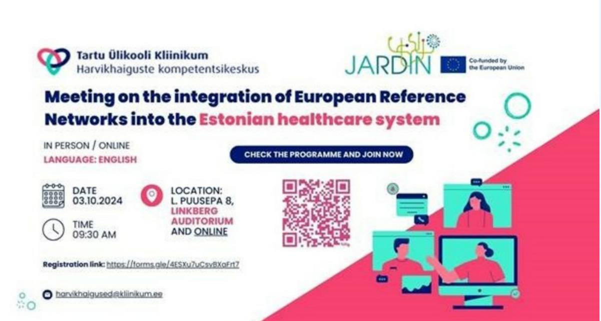
ERN GENTURIS Publications

ERN GENTURIS Publication for **Thematic Group 2 - Adenomatous polyposis syndromes:**

- Large-scale application of ClinGen-InSiGHT APC-specific ACMG/AMP variant classification criteria leads to substantial reduction in VUS, published on 1 October 2024 in AJHG

publications

JARDIN - Integration of ERNs into the Estonian healthcare system



Tartu Ülikooli Kliinikum
Harvikhaiguste kompetentsikeskus

JARDIN

Co-funded by the European Union

Meeting on the integration of European Reference Networks into the Estonian healthcare system

IN PERSON / ONLINE
LANGUAGE: ENGLISH

CHECK THE PROGRAMME AND JOIN NOW

DATE
03.10.2024

TIME
09:30 AM

LOCATION:
L. PUUSEPA 8,
LINKBERG AUDITORIUM
AND ONLINE

Registration link: https://forms.gle/4ESXu7uCsyBXqFrt7

harvikhaigused@kliinikum.ee

On October 3rd, a meeting on the integration of European Reference Networks (ERNs) into the Estonian healthcare system was held at Tartu University Hospital, Estonia. The meeting was initiated by Prof. Katrin Õunap as part of the Joint Action created to achieve the integration of European Reference Networks (ERNs) into National Healthcare Systems (JARDIN).

The key-note speaker, Prof. Alain Verloes, coordinator of **ERN ITHACA**, working at L`Hopital Robert Debre in Paris, shared the experience of his ERN at the EU level.

Ass. Prof. Tiina Kahre, national coordinator of **ERN GENTURIS** in Estonia, presented the work and training possibilities offered by ERN GENTURIS.

Estonia is the smallest of the European countries, thus facing unique challenges in the healthcare system.

Watch the conference (English):

here

Education opportunities

ERN GENTURIS educational webinars

20 November 2024

Molecular profiles of noncanonical malignancies in patients with Lynch or

a BRCA1/2 pathogenic variant

Marjolijn Ligtenberg

11 December 2024

Fertility preserving measures in hereditary cancer syndromes – the oncofertility perspective

Kenny A. Rodriguez-Wallberg

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 course runs from **ctober 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.

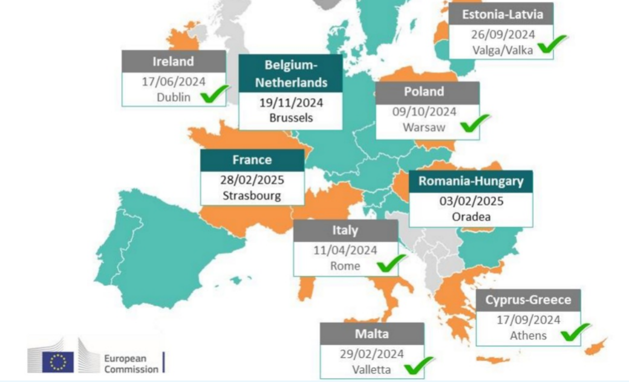
The MOOC is continuously open for enrollment and the access to the content is free for the first 5 weeks.

For more information check the link

course

Workshop on patients’ rights in cross-border healthcare and European Reference Networks

National workshops
Finland
22/10/2024
Helsinki



The next workshop on patients' rights in cross-border healthcare and European Reference Networks will be held in **Brussels, on 19 November.** Working language is Dutch with translation available in English and French, and you can attend it online or on-site.

Please register through the following link:

register

GENERAL NEWS

ERDERA

European Rare Diseases

Research Alliance

ERDERA opens a new era for rare disease research in Europe and beyond. EU-funded ERDERA, European Partnership on Rare Diseases under EU Framework Programme on Research & Innovation Horizon EU and supported by over 170 organizations are uniting to make Europe a leader in rare disease research and innovation.

The kick-off meeting took place on 28-29 October 2024 in Paris, France. Several inspiring speakers addressed the public explaining why rare diseases research will become a top priority for the EU researchers and how rare disease patients can gain from the ERDERA.

This flagship initiative for rare disease community will:

- Unify knowledge, resources and expertise
- Boost clinical research with and for patients
- Spur innovation and EU competitiveness

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".***

website

European Commission news

Health Technology Assessment

The European Commission adopted an Implementing Regulation that sets out the rules for cooperation with the European Medicines Agency in the joint work carried out under the Health Technology Assessment (HTA) Regulation.

These rules define the information to be shared:

- for planning and forecasting of joint clinical assessments and joint scientific consultations;
- for identifying patients, clinical experts and other relevant experts to be involved in these joint procedures; and
- on general scientific and technical matters related to health technology assessment.

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
#HaDEA #EU4Health #HealthUnion
European Commission
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Images source: iStock.com

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

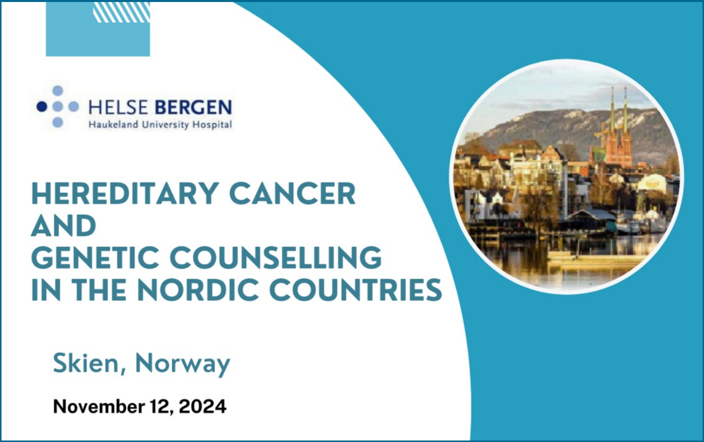
Funding opportunities

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

Hereditary cancer and genetic counselling



HELSE BERGEN
Haukeland University Hospital

HEREDITARY CANCER
AND
GENETIC COUNSELLING
IN THE NORDIC COUNTRIES

Skien, Norway

November 12, 2024

The first meeting about **Hereditary cancer and genetic counselling in the Nordic countries** will take place on **November 12th** in Skien, Norway. The course is open to all healthcare professionals working with hereditary cancer.

registration

ERN GENTURIS GENERAL ANNUAL MEETING

The ERN GENTURIS Annual meeting will take place on 28-29 November 2024 in the Het Trippenhuis, Royal Netherlands Academy of Arts and Sciences, Amsterdam, the Netherlands. Nearly 70 ERN GENTURIS members and patient representatives will participate in this meeting.

For an EU commitment to tackle rare

diseases

For an EU commitment to tackle rare diseases
Az EU elkötelezettsége a ritka betegségek elleni küzdelem mellett
29.11.2024 | Budapest | #RareDiseases
European Economic and Social Committee
HU24EU

The European Economic and Social Committee (EESC), under the auspices of the Hungarian Presidency of the EU Council, is organising a major conference entitled "For an EU commitment to tackle rare diseases" on 29 November 2024 from 9:00 am to 4:30 pm.

more information

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.

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

More information

register here

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Website and LinkedIn

ERN GENTURIS

With every diagnosis we can help an entire family

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