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ERN GENTURIS
Newsletter

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

September 2024

ERN GENTURIS news

5th course in Hereditary Cancer Genetics

77 students from 23 EU member states, Norway, the UK and 4 other non-EU countries participated in the **5th Course in Hereditary Cancer Genetics** in Bertinoro, Italy on 17-20 September 2024. Prof. Nicoline Hoogerbrugge and Dr. Daniela Turchetti chaired the course. The top EU specialists, members of **ERN GENTURIS**, prepared the lectures.

The participants could learn about i.a. the genetic mechanisms of cancer, heritability of cancer and life-time risk, genetic counselling, various tumour syndromes, and more.
Check the programme for more information.

This course aims at delivering up-to-date knowledge on hereditary cancers to clinical and molecular geneticists in training or certified. It creates the best opportunity for interaction and discussion with experts from all over Europe.

ERN GENTURIS 'In the Spotlights' - Dr. Angela Mastronuzzi

Dr. Angela Mastronuzzi, Head of Neuro-Oncology Unit, Area of Hematology-Oncology, Cell and Gene Therapies, and

ERN GENTURIS 'In the Spotlights' - Dr. Kleoniki Roka

Dr. Kleoniki Roka working in the Division of Paediatric Haematology-Oncology, First Department of Paediatrics,

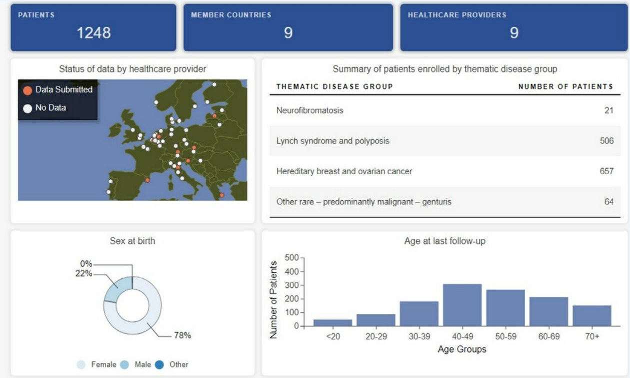
Hematopoietic Transplantation at Ospedale Pediatrico Bambino Gesù in Rome, Italy.
She is a pediatric hematologist-oncologist with a particular focus on central nervous system tumors and cancer predisposing syndromes, implementing molecular diagnostics to study genetic anomalies both at the germline and somatic levels.

read the interview

National & Kapodistrian University of Athens in “Aghia Sophia” Children’s Hospital, Greece.
Together with the other clinicains she organized the first NF Multidisciplinary Team (MDT) Clinic in Greece. The NF MDT Clinic is attending more than 200 children with NF1.

read the interview

ERN GENTURIS Registry



PATIENTS	MEMBER COUNTRIES	HEALTHCARE PROVIDERS
1248	9	9

Status of data by healthcare provider

Summary of patients enrolled by thematic disease group

THEMATIC DISEASE GROUP	NUMBER OF PATIENTS
Neurofibromatosis	21
Lynch syndrome and polyposis	506
Hereditary breast and ovarian cancer	657
Other rare – predominantly malignant – genturis	64

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).

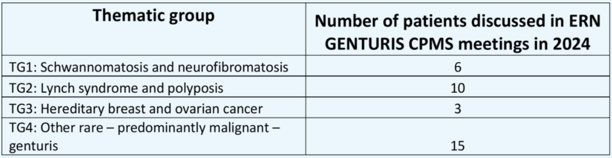
The registry enables a new wave of **research opportunities** from translational to epidemiological studies to clinical trials, and will accelerate the generation of new knowledge on genetic tumour risk syndromes.

- Since the last newsletter (August 2024) more than **155** 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 overview of patients discussed in the CPMS in 2024



Thematic group	Number of patients discussed in ERN GENTURIS CPMS meetings in 2024
TG1: Schwannomatosis and neurofibromatosis	6
TG2: Lynch syndrome and polyposis	10
TG3: Hereditary breast and ovarian cancer	3
TG4: Other rare – predominantly malignant – genturis	15

In the discussions organized through the Clinical Patient Management System (CPMS), ERN GENTURIS has provided tailored expert advice to **34** 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

Please visit our website for a detailed overview of all information regarding the newly developed CPMS (CPMS 2.0). If you are willing to help test CPMS 2.0, please follow the instructions included on the CPMS 2.0 information webpage linked above.

Clinical practice guidelines endorsed by ERN GENTURIS

Endorsed by ERN GENTURIS guidelines for **Thematic Group 3 - hereditary breast and ovarian cancer syndrome (HBOC):**

- **EMQN best practice guidelines for genetic testing in hereditary breast and ovarian cancer** by Trudi McDevitt et al., published in the European Journal of Human Genetics.

endorsed guidelines

Education opportunities

ERN GENTURIS educational webinars

23 October 2024
Adjuvant therapy options for hereditary breast cancer
Tjoung-Won Park-Simon

20 November 2024
Lynch and BRCA1/2 treatment for uncommon cancer types (pending)
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, it aims at delivering up-to-date knowledge on hereditary cancers to clinical and molecular geneticists in training or certified. Opens on **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

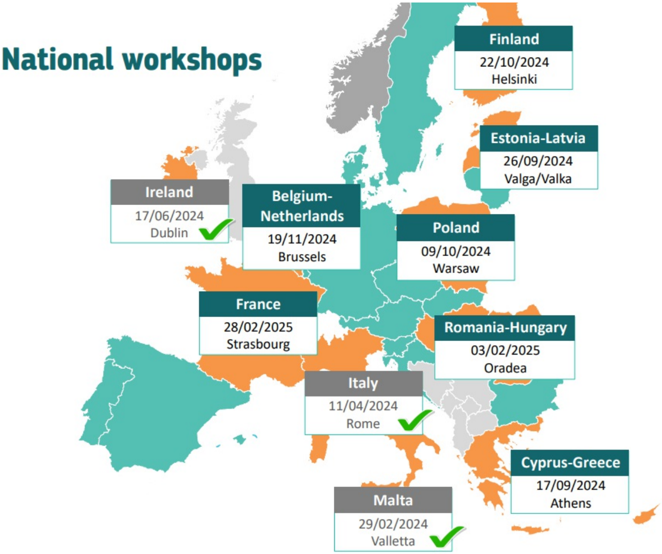
In the context of the European Joint Programme for Rare Diseases EJPRD, co-developed by Chrystelle Colas (ERN GENTURIS), Laurence Faivre (ERN ITHACA), and the Foundation For Rare Diseases, the Massive Open Online Course (MOOC) addresses the diagnostic research progress, types of genetic tests for rare 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

Patients’ rights in cross-border healthcare and European Reference Networks



The next workshop **on patients' rights in cross-border healthcare and European Reference Networks** will be held in **Warsaw, Poland on 9 October.**
Register through the following **link**. You can find the agenda when accessing the registration form.
The workshop will be in Polish, and you have the option to attend it online.

The next workshop will be held in **Helsinki, Finland, on 22 October**.

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.

Read the ERDERA’s press release to learn more:
(...) "With an estimated overall budget of 380 million euros until 2031, ERDERA aims to have a major impact on rare diseases by supporting patient driven research to develop new treatments and diagnostic pathways and harnessing the potential of health and research data, Artificial Intelligence (AI) and digital technologies. (...)"

website

Funding opportunities

European Commission Calls

- HaDEA Calls for **Proposals on Health**
- HaDEA Calls for **Tenders on Health**
- Horizon Europe calls for **Funding on Health**

The info session on the EU4Health

call for proposals

INFO SESSION
EU4Health
2024 open calls
Second wave
RECORDING AND
PRESENTATIONS
AVAILABLE
#HaDEA #EU4Health #HealthUnion #HERA
European
Commission
EU4Health Annual Work Programmes
AWP 2024
AWP 2023
AWP 2022
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This webinar presented the new EU4Health call for proposals with topics on cancer, health promotion and disease prevention, digital health and other actions, funded under the 2024 Annual Work Programme.

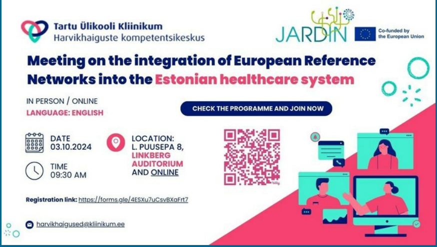
Check out the presentations and watch the recording of the info session:

here

Upcoming Meetings & Events

JARDIN Joint Action in Estonia

Tartu Ülikooli Kliinikum, the Competence Centre of Rare Diseases at Tartu University Hospital is organizing a seminar on the integration of European Reference Networks into the Estonian healthcare system on 3rd October 2024.



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/4ESXu7uCsvBXaFrt7

harvikhaigused@kliinikum.ee

The vice-chair of the **ERN GENTURIS National Coordinators Board, Ass. Prof. Tiina Kahre,** will present the experience of ERN GENTURIS collaboration at the Health Care Provider level.

This seminar will take place within the framework of JARDIN Joint Action. The language will be English, and it is possible to attend in person or online.

Programme

register

Hereditary cancer and genetic counselling

HELSE BERGEN
Haukeland University Hospital

HEREDITARY CANCER AND GENETIC COUNSELLING IN THE NORDIC COUNTRIES

Skien, Norway

November 12, 2024

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.

Deadline for registration is October 22, 2024 .

Abstracts:
You can choose between two categories:

1. Scientific project: submit a structured abstract describing your study. The abstract should include names of all authors and their affiliations. The name of the person presenting the abstract must be underlined. Font: Times New Roman, size: 12; Maximum length: 250 words (excluding title, authors, and affiliations).

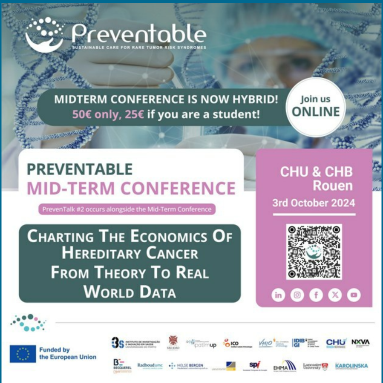
2. Challenging clinical case: submit a PowerPoint presentation on a challenging case you want to discuss with our Nordic expert panel. The presentation should be of maximum 2-3 slides (1 slide pedigree, 1-2 slides pertinent details). In the e-mail you should include a title of your presentation, your name, place of work, and position. Junior colleagues will be prioritized for the case presentations.

Send your abstract/PPT to: cathrine.bjorvatn@helse-bergen.no
Deadline for abstract submission is October 1st .

registration

Charting the Economics of Hereditary Cancer: From Theory to Real World Data

The French team of the PREVENTABLE project, Centre Hospitalier Universitaire de Rouen and Centre Regional de Lutte Contre Le Cancer Henri Becquerel Rouen, is organising a Midterm Conference on October 3, 2024 in Rouen, France. During the conference four distinguished keynote speakers, in the areas of Tumor Risk Syndromes, health economics, and communication in healthcare, will present their work.



Preventable
SUSTAINABLE CARE FOR RARE TUMOR RISK SYNDROMES

MIDTERM CONFERENCE IS NOW HYBRID!
50€ only, 25€ if you are a student!

Join us ONLINE

PREVENTABLE
MID-TERM CONFERENCE
PrevenTalk #2 occurs alongside the Mid-Term Conference

Charting The Economics Of Hereditary Cancer From Theory To Real World Data

CHU & CHB Rouen
3rd October 2024

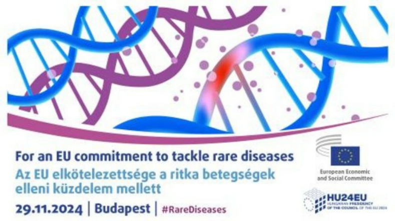
Funded by the European Union

Agenda: LINK

more info

For an EU commitment to tackle rare diseases

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.



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

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

Call for Abstracts related to 'ERN Research' deadline 1st October 2024

register here

Visit our:
Website and LinkedIn

ERN GENTURIS

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

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