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ERN GENTURIS Newsletter

European Reference Network
Genetic Tumour Risk Syndromes (ERN GENTURIS)

June 2024

ERN GENTURIS news

ERN GENTURIS meeting in Berlin

The **ERN GENTURIS meeting** took place prior to the ESHG conference in Berlin on 31 May and 1 June 2024. More than 50 ERN GENTURIS members participated in the meeting.

The ePAG representative **Darja Molan** from Europa Donna Slovenia, presented on behalf of the **ERN GENTURIS ePAGs.** She was diagnosed as a BRCA1 carrier with breast cancer 11 years ago, at age 30. She shared that healthcare systems currently focus on disease management, rather than disease prevention; there should be more attention and research funding for prevention of hereditary cancers.

Darja encouraged the ERN GENTURIS members to write proposals for the open HORIZON-MISS-2024-CANCER-01-03 call on Accessible and affordable tests to advance early detection of heritable cancers in European regions.

EURACAN and ERN GENTURIS: training session in Finland

ERN EURACAN AND GENTURIS:
Rare Cancers and Hereditary Cancer Syndromes educational symposium
Helsinki, 24.5.2024

Hereditary tumor syndromes:
diagnosis, network structures,
and variant classification

Stefan Aretz
Institute of Human Genetics
Center for Hereditary Tumour Syndromes
ERN GENTURIS
University Hospital Bonn
Germany

CIO
ukb universitäts klinikumbonn
Zentrum für seltene Erkrankungen Bonn
European Reference Network
DRNETS
Deutsches Konsortium Familiärer Darmkrebs

On 24th May 2024, took place a training organized by the Southern Cancer Center and the Sisä-Suomen Syöpäkeskus in Helsinki, Finland.

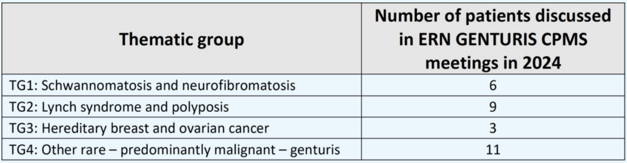
The goals of this training were: strengthening the cooperation of professionals from the **ERN EURACAN and ERN GENTURIS** networks and a joint discussion between patients and healthcare professionals.

The training included two expert lectures:

- **Prof. Stefan Aretz**, ERN GENTURIS (on the picture) about Hereditary tumor syndromes: diagnosis, network structures, and variant classification.
- **Prof. Jean-Yves Blay,** EURACAN about: Genome screening aiming at precision treatment in rare cancers - for who, what tests and when?

The ERN GENTURIS session was chaired by **Maria Haanpää**.
Sirkku Peltonen presented the new NF1 follow-up recommendations, and **Minna Pöyhönen** presented the ERN registries.

ERN GENTURIS overview of patients discussed in the CPMS in 2024

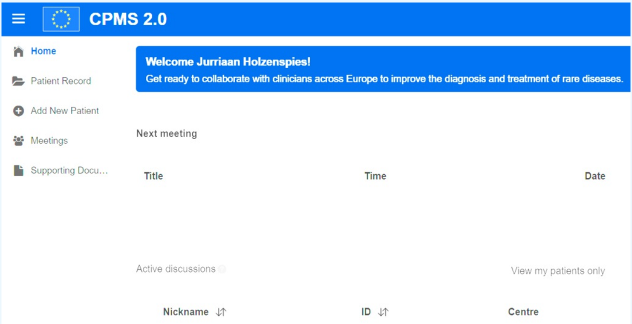


Thematic group	Number of patients discussed in ERN GENTURIS CPMS meetings in 2024
TG1: Schwannomatosis and neurofibromatosis	6
TG2: Lynch syndrome and polyposis	9
TG3: Hereditary breast and ovarian cancer	3
TG4: Other rare – predominantly malignant – genturis	11

In the discussions organized through the Clinical Patient Management System (CPMS), ERN GENTURIS has provided tailored expert advice to **29** 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

Transition to CPMS 2.0



CPMS 2.0

Welcome Jurriaan Holzenspies!

Get ready to collaborate with clinicians across Europe to improve the diagnosis and treatment of rare diseases.

Next meeting

Title	Time	Date

Active discussions

View my patients only

Nickname	ID	Centre

During the summer break, ERN GENTURIS will transition to the **new CPMS** (**CPMS 2.0**). As a member of ERN GENTURIS, you can help ensure a smooth transition to the new system by closing any currently open panels (see Quick guide 3 - Dashboard and panel advancement) and testing the new system over the summer (to volunteer, please contact CPMS helpdesk manager Jurriaan Hölzenspies (here).

CPMS 2.0 transition - overview of the most important information

- No interruption of patient discussions in recurring CPMS meetings – we will continue to use the current method (email announcement in combination with meetings held in GoToMeeting) until we have confirmed that our recurring meetings can be held in the new CPMS. Once confirmed our entire network will be informed and our meetings will be held in the new CPMS from then on.
- All user data will be migrated during the transition, so your account will be automatically transferred and you will be able to log in to the

new CPMS immediately after the transfer. You will be informed of the transition date as soon as it has been selected.

- All patient data will be migrated, but it may not be possible to do this at the same time as we start using the new CPMS, because the data migration process is very complex. If moving the patient data proves to be problematic, we will start using the new CPMS without migration of patient data. The patient data from the old CPMS will then be moved to the new CPMS at a later time point.

Education opportunities

ERN GENTURIS educational webinars

25 September 2024
ERN GENTURIS BHD syndrome guidelines
Marianne Geilswijk

More on our website:

webinars

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

European Reference Networks at the ESHG 2024

12:15 - 13:15 Get2gether European Reference Networks

CHAIRS: PROF. NICOLINE HOOGERBRUGGE, PROF. CARLA OLIVEIRA

Pathway development within ERNs – ERN-RND

European Reference Network | Neurological Diseases (ERN-RND)

Patient journey – Friedreich's Ataxia

Hélène Dollfus

Rare diseases guidelines and associated documents: the impact of ERNs

024 Berlin

European Reference Networks

JUNE 1–4, 2024

Room A4

First Symptoms
Diagnosis
Treatment
Monitoring
Disease
Clinic
Challenges
Goals
ESHG

On Sunday, June 2 2024 at the European Human Genetics Conference (ESHG) in Berlin took place the **Get2gether European Reference Networks** aimed at presenting and raising awareness about the presence and the usefulness of the **ERNs** and at promoting networking and professional connections among those clinically and academically interested in rare diseases. Attendees could learn about:

1. Integration of ERNs into the national healthcare systems
2. Implementation of ERNs guidelines in the national healthcare system
3. Transversal use of ERN webinars by geneticists

The chairs of the session were: **Prof. Carla Oliveira** and **Prof. Nicoline Hoogerbrugge** (**ERN GENTURIS**), and the speakers **Prof. Hélène Dollfus** (**ERN-EYE)** and **Dr. Holm Graessner** (**ERN-RND**).

The four ERNs: **ERN-EYE, ERN Euro-NMD, ERN ITHACA and ERN GENTURIS** shared a booth to raise awareness about the Networks, our work, goals and mission.

ESHG 2024 ERNs booth

European Reference Networks
864

European Reference Networks
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European Reference Networks
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European Reference Networks
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Pre-InSiGHT meeting for patients and patient representatives with Lynch syndrome or polyposis

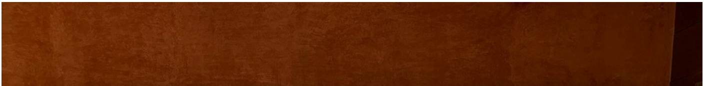
On the evening of Tuesday June 18 2024, the pre-InSiGHT meeting for patients and patient representatives with **Lynch syndrome or polyposis** was broadcast to a live online audience from Barcelona, Spain. This meeting provided a unique opportunity for **lynch and polyposis** patients and their families to obtain clear and accurate information about this disease from medical experts, including the possibility to ask them questions directly (see the picture of the live questions session below).

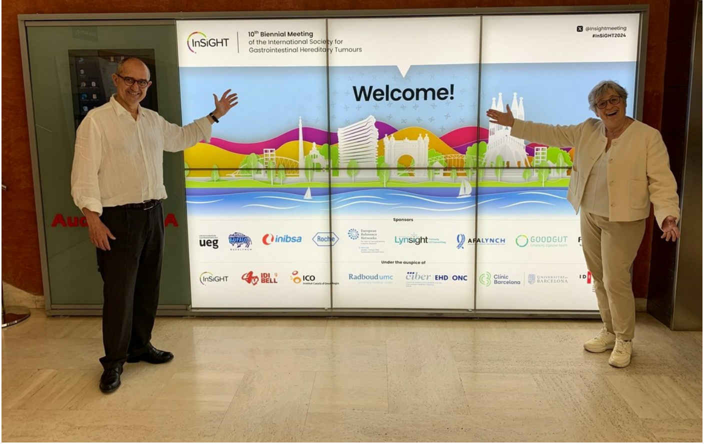
The meeting was a great success and patients were very enthusiastic about the quality of the meeting and the opportunity to ask a panel of experts specific questions about **Lynch syndrome and polyposis**.

Screenshot of the “Pre-InSiGHT meeting for patients and patient representatives with Lynch syndrome or polyposis” questions session. From left to right and top to bottom, **Dr. Judith Balmaña, Prof. Nicoline Hoogerbrugge, Dr. Toni Seppälä, Dr. Gabriel Capellá, Brandie Leach and Dr. Francesc Balaguer**.

For more information and the meeting programme:

programme



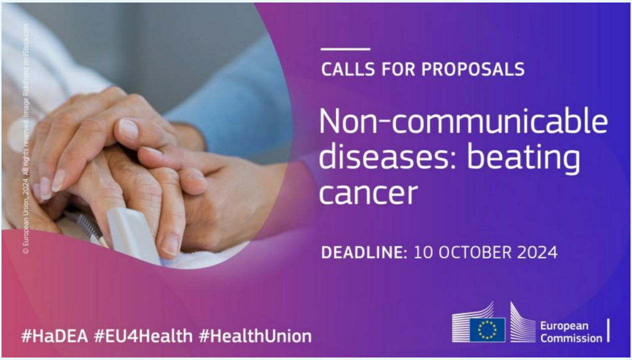

InSiGHT
10th Biennial Meeting of the International Society for Gastrointestinal Hereditary Tumours
@insightmeeting #InSiGHT2024
Welcome!
Sponsors
Under the auspice of

Funding opportunities

European Commission Calls

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

EU4Health call for proposals on cancer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CALLS FOR PROPOSALS
Non-communicable diseases: beating cancer
DEADLINE: 10 OCTOBER 2024
#HaDEA #EU4Health #HealthUnion
European Commission

HaDEA has published the call for proposals on cancer under the 2024 EU4Health work programme:

- EU4H-2024-PJ-02-1: Call for proposals to support integration of cancer images into the federated pan-European infrastructure to foster screening programmes - CR-g-24-37
- EU4H-2024-PJ-02-2: Call for proposals to increase health literacy for cancer prevention and care - CR-g-24-39
- EU4H-2024-PJ-02-3: Call for proposals on the development of social services for psychosocial support and rehabilitation for children and their families in paediatric oncology clinics in Member States and countries associated to the EU4Health Programme - CR-g-24-43
- EU4H-2024-PJ-02-4: Call for proposals on EU Network of Youth Cancer Survivors - CR-g-24-45

more information

Upcoming Meetings & Events

5th course in Hereditary Cancer Genetics

This course, organized by members of **ERN GENTURIS**, 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, in the fabulous environment of Bertinoro, the headquarters of the ESHG sponsored courses. The faculty combines experts from many fields of cancer genetics known for their didactic skills. Participants are encouraged to present a clinical or genetic case in a Poster format for on-site discussion.
The course will take place on 17-20 September 2024.

more information

ERICA GA & ERN Research Conference

Save the Date:
ERICA General Assembly &
ERN Research Conference
11-13 December 2024 in Udine, Italy
ERICA
European Reference Network
MetabERN

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

This event is hosted by MetabERN.

There will also be a Call on Abstracts related to ERN Research.

more information

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ERN GENTURIS
With every diagnosis we can help an entire family

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