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ERN GENTURIS Newsletter

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

February 2025

ERN GENTURIS news

Rare Disease Day 2025 ERN GENTURIS

Ghent, BE
Nijmegen, NL
Rome, IT
Tartu, EE
RARE DISEASE DAY
SUPPORTED BY
ERN GENTURIS MEMBERS

Athens, EL
Erlangen, DE
Porto, PT

The ERN GENTURIS members have united in support of **Rare Disease Day, February 28, 2025**. Together, we raise awareness and advocate for those impacted by genetic tumour risk syndromes. Check our news & events page.

Genturis patients are at very high hereditary risk of developing common cancers, which are often located in multiple organ systems. In case they are diagnosed with cancer they may need different treatment and follow-up as compared to non-hereditary cancers.

- For information about ERN GENTURIS, see: website
- For information on how to participate in genturis research, see: research projects
- Information for health professionals and patients considering a genetic test to look for an inherited cause of cancer, see: cancer genetics

European Reference Networks unite for Rare Disease Day

European Reference Networks

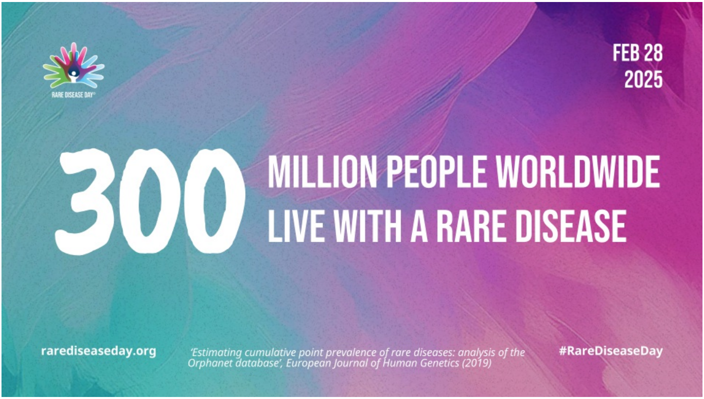
Rare patients.
Rare experts.
Real outcomes.
https://.health.ec.europa.eu//rare-diseases-and-european-reference-networks
European Reference Networks
Funded by the European Union

This year, the 24 European Reference Networks (ERNs) united to mark **Rare Disease Day 28 February 2025**, launching a shared communication strategy. The initiative aims to raise awareness among healthcare providers and the general public, promoting a better understanding of the ERNs and their role in improving care for rare diseases.

The European Commission published, on the occasion of Rare Disease Day, a booklet celebrating the dedication and expertise of the European Reference Networks, which make a tangible difference in the lives of patients with rare diseases and their families, as well as two new factsheets.

Please find below further details for your information:
1. Factsheet: "EU delivering on rare diseases for patients and families", 4 pages. Available in EN/FR/DE/IT on the ERNs
2. Factsheet: "How the European Commission works on Rare Diseases and the European Reference Networks (ERNs)", 3 pages. Available in EN/FR/DE/IT on the ERNs
3. Booklet: "European Reference Networks. A success story for patients living with a rare disease", 64 pages. Available in EN

european reference networks


RARE DISEASE DAY
FEB 28
2025
300 MILLION PEOPLE WORLDWIDE
LIVE WITH A RARE DISEASE
rarediseaseday.org
'Estimating cumulative point prevalence of rare diseases: analysis of the Orphanet database', European Journal of Human Genetics (2019)
#RareDiseaseDay

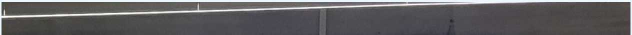
In the Spotlights interviews

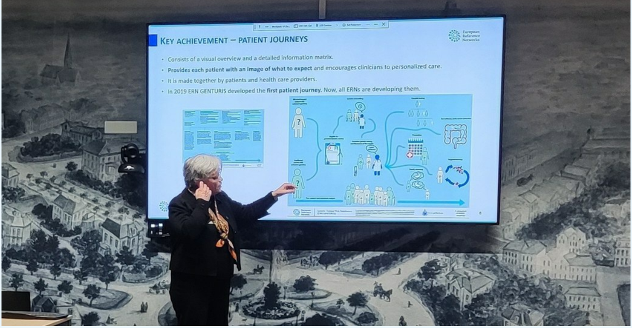
In our newest 'Spotlights,' you can read interviews with Task Force 6 Chair **Liliana Sousa** (Junior Researcher – Project Manager) and TF6 member **Mikk Tooming** (Clinical Molecular Geneticist - Research & Development).

You can learn how ERN GENTURIS helps professionals collaborate and improve patient care, as well as which initiative within ERN GENTURIS has had the most impact from a project management perspective.

in the spotlights

Nicoline Hoogerbrugge's presentation for Dutch Ministry of VWS





KEY ACHIEVEMENT – PATIENT JOURNEYS

- Consists of a visual overview and a detailed information matrix.
- **Provides each patient with an image of what to expect** and encourages clinicians to personalized care.
- It is made together by patients and health care providers.
- In 2019 ERN GENTURIS developed the **first patient journey.** Now, all ERNs are developing them.

A delegation of the Dutch Ministry of Health, Welfare and Sport (VWS) visited Radboudumc on Monday 17 February.

Representatives of the Netherlands Federation of University Medical Centres (NFU - Nederlandse Federatie van Universitair Medische Centra -NFU) and of the Dutch Expertise Centers for Rare Disorders (Expertisecentrum Zeldzame Aandoeningen - ECZA) and European Reference Networks (ERNs) informed the delegation about the importance of combining knowledge and skills about the rare diseases in expertise centers and ERNs to ensure better and faster treatment.

Professor Nicoline Hoogerbrugge (van der Linden), coordinator of ERN GENTURIS, presented key actions and output of ERN GENTURIS.

It is a great example of dissemination activities at the national level to spread knowledge about the work done by the ERNs.

CMMRD guideline presentation

On January 29th the 7th workshop of the European consortium Care for Constitutional Mismatch Repair deficiency (C4CMMRD) took place at the Inselspital, Bern, Switzerland.

This workshop held in tandem with the SIOPE Host Genome WG meeting brought together 95 (47 in persons and 48 online) clinical and moleculargeneticists, paediatric oncologists and researchers focussing on rare childhood cancer predisposition syndrome and related disorders.

This meeting was an ideal place to present the newly approved and published ERN GENTURIS guidelines on CMMRD diagnosis, genetic counselling, surveillance, quality of life, and clinical management. The meeting was organised by Katharina Wimmer, Chrystelle Colas and Léa Guerrini-Rousseau.

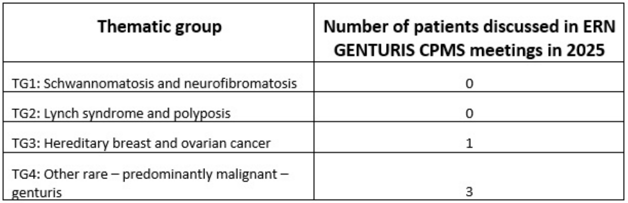
Link: CMMRD guideline

Impact of Rare Conditions on Mental Health & Relationships by Matt Bolz-Johnson

EURORDIS
Impact of Rare Conditions on Mental Health & Relationships
Matt Bolz-Johnson, Mental Health Lead, EURORDIS

Matt Bolz-Johnson, Mental Health Lead & Healthcare Advisor at EURORDIS, recently addressed clinical geneticists in training at Radboudumc and stressed the significant impact rare conditions have on both mental health and relationships. He highlighted that individuals living with rare diseases often face increased levels of anxiety, depression, and emotional distress due to the challenges of diagnosis, treatment, and a lack of societal awareness.

ERN GENTURIS CPMS news



Thematic group	Number of patients discussed in ERN GENTURIS CPMS meetings in 2025
TG1: Schwannomatosis and neurofibromatosis	0
TG2: Lynch syndrome and polyposis	0
TG3: Hereditary breast and ovarian cancer	1
TG4: Other rare – predominantly malignant – genturis	3

ERN GENTURIS overview of patients in 2025

In the discussions organized through the Clinical Patient Management System (CPMS 2.0), ERN GENTURIS has provided tailored expert advice to **4** genturis patients and their families in 2025 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
- Guides on how to use CPMS 2.0

CPMS 2.0 information for members

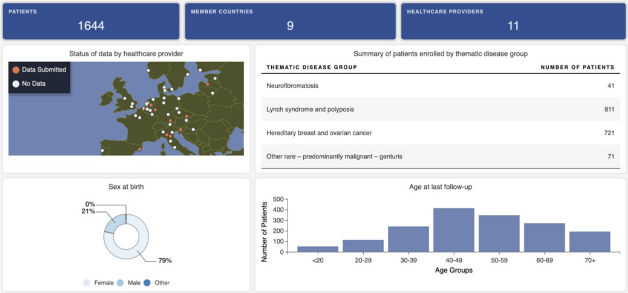
CPMS recurring meeting connectivity

Several of our members have experienced problems connecting to our CPMS recurring meetings using the CPMS 2.0 built-in video conferencing tool. As these connectivity problems are almost exclusively related to the firewall settings of the institute that the user is trying to connect from, it can usually be solved by switching to a different internet connection. For more permanent potential solutions for connectivity issues, please see below.
If you are experiencing connectivity issues please follow these steps:

1. Notify our CPMS helpdesk manager, Jurriaan Hölzenspies
2. Ask your IT department to unblock the CPMS domain (cpms2.ern-net.eu) and please make sure to mention that this is for a video conferencing tool. If needed, you can ask them to help you with the test in step 3 as well.
3. To test your connectivity after the CPMS has been unblocked in step 2, go to cpms2.ern-net.eu, log in, navigate to “Meetings” in the menu on the left and then click “Join” next to any of the listed meetings to test if you receive the popup to ask for permission to use your audio/video. Clicking the join button and opening the meeting does not affect the meeting in any way and it can still be opened again on the scheduled date/time.

A more detailed troubleshooting guide for CPMS meeting connectivity issues will be published on the genturis website in the next week on our CPMS guides and videos page.

ERN GENTURIS Registry



PATIENTS 1644

MEMBER COUNTRIES 9

HEALTHCARE PROVIDERS 11

Summary of patients enrolled by thematic disease group

THEMATIC DISEASE GROUP	NUMBER OF PATIENTS
Neurofibromatosis	41
Lynch syndrome and polyposis	811
Hereditary breast and ovarian cancer	721
Other rare – predominantly malignant – genturis	71

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 our last mailing , more than 396 new patients have been included in the GENTURIS registry.
- Recently two HCPs have started to enroll patients: Azienda Ospedaliero-Universitaria di Parma and Center for Hereditary Tumor Syndromes (CHT), University Hospital Bonn.

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

more information

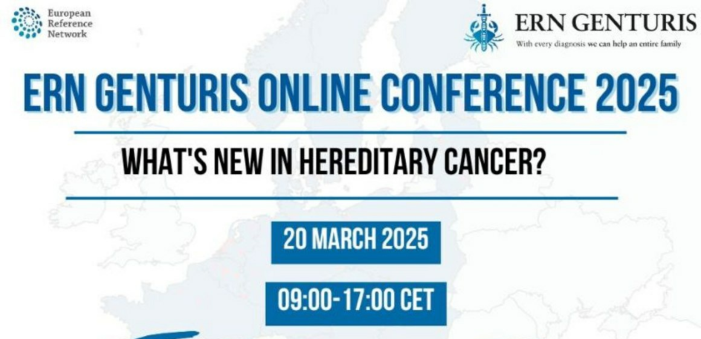
Education opportunities

ERN GENTURIS educational webinars

12 March 2025
Genetic cancer predisposition in adolescents and young adults
Richarda de Voer

Watch the previous webinars here:

webinars

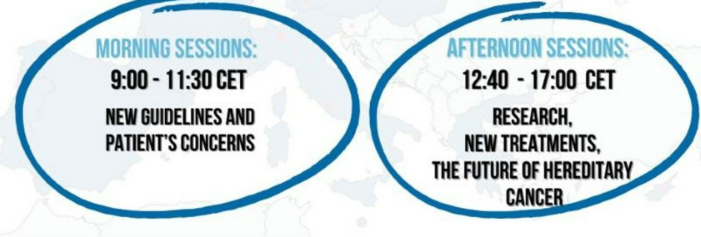

European Reference Network
ERN GENTURIS
With every diagnosis we can help an entire family

ERN GENTURIS ONLINE CONFERENCE 2025

WHAT'S NEW IN HEREDITARY CANCER?

20 MARCH 2025

09:00-17:00 CET



MORNING SESSIONS:
9:00 - 11:30 CET
NEW GUIDELINES AND PATIENT'S CONCERNS

AFTERNOON SESSIONS:
12:40 - 17:00 CET
RESEARCH,
NEW TREATMENTS,
THE FUTURE OF HEREDITARY CANCER

A printable pdf version of the programme is available here.

Free registration & more information:

conference

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

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

Conference on Rare Diseases in Warsaw, Poland

The Ministry of Health of Poland, in cooperation with the European Economic and Social Committee (EESC) and Medical University of Warsaw, would like to announce a Conference on Rare Diseases will take place on **10th of April 2025**, in Warsaw.

The conference will be followed by a side-event, held on **11th of April 2025**, dedicated specifically to clinicians, researchers and patients.

More information here: [Conference on 'Towards an EU action plan on rare diseases' | EESC]

European Human Genetics Conference ESHG 2025

ESHG2025
EUROPEAN HUMAN GENETICS CONFERENCE
24-27 May | Allianz MiCo, Milan | Italy
ESHG

The European Human Genetics Conference will take place on **24-27 May 2025** in Milan, Italy.

more information

Visit our:
Website and LinkedIn

ERN GENTURIS
With every diagnosis we can help an entire family

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