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ERN GENTURIS Newsletter

Genetic Tumour Risk Syndromes (ERN GENTURIS)

January 2025

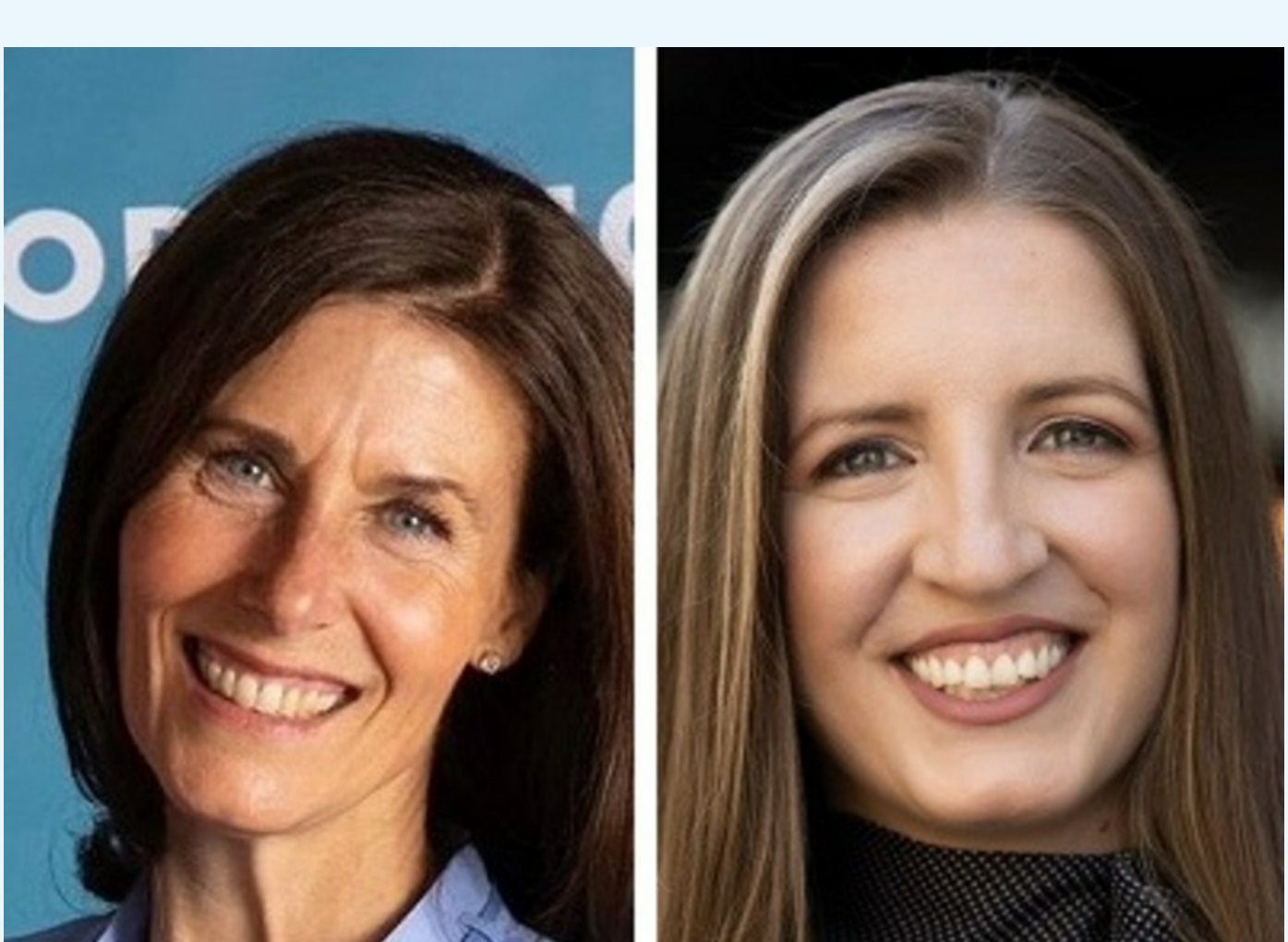
ERN GENTURIS news

A printable pdf version of the programme is available here.

Free registration & more information:

conference

In the Spotlights - ePAGs



Georgina Hoffmann and Joana Pais, our two new ERN GENTURIS Patient Representatives, shared in the interview what it took to understand their genetic risk, how cancer has impacted them and their family members, and what it's like living with rare genetic tumour syndromes. They also shared a very important message for the international medical community.

in the spotlights

Solve-RD manuscript

The newly published Solve-RD's manuscript based on the years of work in Nature Medicine: **"Genomic Reanalysis of a Pan-European Rare Disease Resource Yields New Diagnoses"**.

More than 500 European patients with unknown conditions have received a diagnosis through new genetic research. This includes patients with rare neurological disorders, severe intellectual disabilities, muscle diseases, and hereditary gastrointestinal cancer. These diagnoses were achieved through extensive European collaboration of 300 experts from 12 European countries, led by researchers from the University of Tübingen, Radboudumc, and the National Center for Genomic Analysis in Barcelona.

Congratulations to all authors, all researchers and particularly to the 4 ERNs:

- ERN ITHACA
- ERN GENTURIS
- EURO-NMD European Reference Network for Rare Neuromuscular Diseases
- ERN-RND, European Reference Network for Rare Neurological Diseases

manuscript

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	0
TG4: Other rare – predominantly malignant – genturis	1

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 **1** genturis patients and their families in 2025.
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

>>Important information regarding ERN GENTURIS CPMS meetings<<

All CPMS meetings will take place inside CPMS 2.0 from now on

- **CPMS 2.0 (https://cpms2.ern-net.eu/)**
  - !!!Please note: Patients that were already uploaded in the old CPMS should not be entered into the new CPMS. These cases will become available for discussion in CPMS 2.0 after the patient data migration (will be scheduled before the end of January 2025).
  - To schedule a meeting for your patient, please contact the ERN GENTURIS CPMS helpdesk manager Jurriaan Hölzenspies (send an email to jurriaan.holzenspies@radboudumc.nl or use the chat function in CPMS 2.0 at the top right of the page to contact him).
  - To submit patients for discussion, an EU login account is required. To create an account and request access to the ERN GENTURIS CPMS, please follow these steps:
    - Go to https://webgate.ec.europa.eu/cas/eim/external/register.cgi and follow the on-screen instructions
    - When the account from the previous step is active, log in at https://cpms2.ern-net.eu/ to request access to the GENTURIS CPMS

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

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

ERICA ERN Research Conference

The ERICA ERN Research Conference took place from 11-13 December 2024 in Udine, Italy and was hosted by MetabERN.

ERICA aims to set the standards for future rare disease programmes like the European Rare Diseases Research Alliance (ERDERA).

Check the presentations, posters, and event photos here:

website

European Commission news

Public consultation

TEHDAS2 joint action has launched a 30-day public consultation on four draft guidelines and technical specifications designed to support the effective secondary use of health data across Europe as part of the European Health Data Space (EHDS).

The consultation closes on 28 February 2025. Stakeholders are encouraged to review the documents and provide their feedback through dedicated Webropol surveys available for each consultation.

consultation

Commission lanched JANE-2

EU JOINT ACTION JANE NETWORKS OF EXPERTISE ON CANCER

The second Joint Action of the EU Networks of Expertise on Cancer (JANE-2) has been launched on 29 January 2025. This is one of the largest Joint Actions on cancer, supported by EUR 40.5 million in funding from the EU4Health programme. JANE-2 has 121 partners from 29 European countries and is coordinated by Italy.
The objective of the Joint Action is to establish seven **Networks of Expertise on cancer in the EU**, implementing the recommendations of the 2021 Europe's Beating Cancer Plan. The Networks bring together the highest expertise with the view of improving EU cooperation in the fight against cancer.

read more

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

Workshops on Rare Diseases in Cross-Border Healthcare

The conference on Patients' Rights and Rare Diseases in the European Union cross-border healthcare **co-organised by Romania and Hungary** will be held in **Oradea, Romania, on 5 February 2025**. Working language will be in Romanian and Hungarian with translation available in English and Hungarian/Romanian.
There are a few available places left for the on-site participation, register through the following link.

The last workshop on Patients' Rights and Rare Diseases in the European Union cross-border healthcare **organised by France** will take place in Strasbourg **on 27 February 2025.** It will be held in the premises of the European Parliament.

Register under link below:

register

Conference on Rare Diseases in Warsaw, Poland

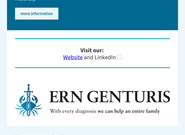
Ministry of Health Republic of Poland | European Economic and Social Committee | Medical University of Warsaw

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 will follow.

European Human Genetics Conference ESHG 2025



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**