ERN GENTURIS Newsletter

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

November 2025

ERN GENTURIS news

ERN GENTURIS annual meeting 2025

The ERN GENTURIS annual meeting took place on 27 and 28 November 2025 in Amsterdam, the Netherlands.

4 Patient Representatives and more than 70 ERN GENTURIS members joined the meeting and shared their experience and expertise.

Discussion focussed on collaboration for the clinical guidelines development, research possibilties, registry and education activities.

ERN GENTURIS Podcast

New ERN GENTURIS Podcast Episode: *The role of genetic counselling: Why it matters?*

In this episode, we explore the crucial role of genetic counselling for families affected by hereditary syndromes. We delve into how genetic counselling helps guide patients and families through complex decisions, provides much-needed psychological support, and ultimately improves long-term health outcomes.

We are joined by two expert guests: **Robin de Putter,** a clinical geneticist and medical oncologist specialising in cancer predisposition at UZ Gent in Belgium and **Luzia Garrido**, a genetic nurse at ULS São João in Porto, Portugal, who offers dedicated support to families throughout their genetic counselling journey. Together, they explain what genetic counselling entails, why it's so important, and how it empowers families to make informed decisions about their health.

This episode is full of insights you won't want to miss!

Listen now and follow the series to stay informed!

spotify

In the spotlights interview

In the latest Spotlight interview, **Carla Oliveira**, a biochemist and leader in

the field of rare tumour risk syndromes at i3S - Instituto de Investigação e Inovação em Saúde (i3S), shares her inspiring journey, from her work in Human Molecular Genetics to her pivotal role in ERN GENTURIS.

As the Institutional Lead of the first GENTURIS reference centre in Portugal and the National Coordinator, Carla has played a key role in improving the care and outcomes for patients with rare genetic tumour risk syndromes.
In this candid interview, she opens up about her passion for research, her leadership in establishing reference centres, and how ERN GENTURIS is transforming the way doctors approach these complex conditions.

interview

ERN GENTURIS CPMS news

Thematic group	Number of patients discussed in ERN GENTURIS CPMS meetings in 2025
TG1: Schwannomatosis and neurofibromatosis	5
TG2: Lynch syndrome and polyposis	11
TG3: Hereditary breast and ovarian cancer	6
TG4: Other rare – predominantly malignant – genturis	19

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 **46** 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 invitations for 2026

The new CPMS recurring meeting invitations for 2026 were sent out to all our CPMS users on Monday 24 November 2025. As these meetings are held separately for TG1 and TG2-4 on the

first and third Friday of the month, you will have received the following four invitations:

- ERN GENTURIS CPMS meeting for **TG1**: Schwannomatosis and neurofibromatosis – **First** Friday of the month
- ERN GENTURIS CPMS meeting for **TG2-4**: Lynch and polyposis, HBOC and rare genturis - **First** Friday of the month
- ERN GENTURIS CPMS meeting for **TG1**: Schwannomatosis and neurofibromatosis - **Third** Friday of the month
- ERN GENTURIS CPMS meeting for **TG2-4**: Lynch and polyposis, HBOC and rare genturis - **Third** Friday of the month

CPMS upcoming change to recurring meetings

As part of an improvement proposed by ERN GENTURIS, the CPMS support team is currently working to implement changes to the recurring meeting system that will allow our clinicians to add their own patients to a meeting. The system will function similarly to the old CPMS in that there will be 6 available 10 min slots per meeting and when a patient is added, they are always entered into the next available slot. When the change has been implemented, you will be notified of the change and the guide for scheduling a meeting on our website will be updated.

CPMS scheduling issues

A recently discovered bug in the CPMS causes the system to send out a cancellation and a new meeting invite each time a participant is added to the meeting. This issue has been reported to the CPMS support team and the developer is working on a solution. You will be informed when the issue has been resolved, but before that happens, you may receive additional calendar cancellations and invitations due to this bug. This can cause additional calendar entries for the same meeting, but until this has been fixed, you can simply delete the extra entries from your calendar if needed. As of November 2025, a fix for newly created meetings was implemented, so this should be fixed in the 2026 meeting series for which you received invitations recently (see above).

CPMS meeting connectivity issues

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 notify our CPMS helpdesk manager, Jurriaan Hölzenspies
A troubleshooting guide for CPMS meeting connectivity issues is available on the genturis website on our CPMS guides and videos page.

4th NATIONAL AIFET CONGRESS

The 4th National AIFET Congress (Italian Association for Cancer Familiarity and Heredity), took place in Milan from November 19 to 21, 2025, at the Fondazione IRCCS Istituto Nazionale dei Tumori. Several members of ERN GENTURIS from all over Italy participated in this meeting and Chystelle Colas working at the Insitut Curie in Paris, France, presented Constitutional Mismatch Repair Deficiency syndrome: Clinical framing, surveillance, and immunotherapy as a new treatment option. ERN GENTURIS Guidelines Recommendations.

more information

Education opportunities

ERN GENTURIS educational webinars

21 January 2026
Hereditary childhood cancer
Marjolijn Jongmans

25 February 2026

DICER1-associated thyroid tumours - the pathologist's perspective
Giovanni Tallini

11 March 2026

PALB2 risk and surveillance
Marc Tischkowitz

24 April 2026

CHEK2 germline pathogenic variants
Richarda de Voer & Marleen Kets

More inforrmation:

webinars

General news

In-depth analysis of the public consultation of the European Parliament on rare diseases

This in-depth analysis presents the findings of the public consultation on rare diseases conducted by the European Parliament in March 2025. Contributions from a wide range of participants, including patients and families, patient support groups, healthcare professionals, stakeholder interest groups and international organisations, government and other public bodies, aim to guide future policy actions in the field of rare diseases. This document was provided by the Policy Department for Transformation, Innovation and Health at the request of the Committee on Public Health.

report

Continuous Monitoring Report for the ERNs

The European Commission has published the first-ever Continuous Monitoring Report for the ERNs, confirming a 160% increase in new patients referred to ERN centres between 2018 and 2024, clear evidence of how this EU infrastructure strengthens Europe's health capacity and resilience. The report presents detailed indicators across seven core areas, including clinical activity, guidelines, registries, training and cross-border collaboration, reflecting the 2023 & 2024 monitoring cycles.

ERNs connect 1,600+ specialised units in 375 hospitals, creating a single pool of rare disease expertise no country could build alone. An estimated 30 million EU citizens are living with a rare disease. That's a huge challenge, and ERNs are working together to make sure everyone gets the specialised care they need.

The report documents over 2,100 virtual CPMS consultations, delivering expert, multidisciplinary advice across borders so patients can access specialist input without the need to travel.

ERNs, demonstrate the power of European cooperation, strengthening health resilience, supporting clinical excellence, and improving access to expertise for rare disease patients across all Member States.

Read the report and learn more about how ERNs and the clinicians and multidisciplinary teams who power them, support patients across Europe.

news

Call for Experts: New EU Panel on Paediatrics and Rare Diseases (Medical Devices)

The European Commission has announced a call for expressions of interest for a newly established expert panel on **Paediatrics and Rare Diseases** in the field of medical devices. The EMA reports that additional applications are urgently needed for this panel.

Colleagues with relevant expertise are encouraged to apply and share this call within their networks.

Applicants should ensure they select “Paediatrics and rare diseases” as one of their chosen panels during the application process. Application guidance is provided in the Annex I – Application Guidance Experts Management Tool of the Call for Expressions of Interest.

more information

Funding opportunities

European Commission Calls

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

ERDERA Networking Support Scheme

Round 2 of ERDERA’s Networking Support Scheme is open!

ERDERA’s Networking Support Scheme is a competitive funding instrument supporting transnational networking events in the field of rare diseases and rare cancers.

Applications are reviewed every six months, and proposal round 2 is now open until **7 April 2026 at 14:00 (CET)**.

The NSS provides funding of up to €30,000 per event to support transnational networking activities that promote knowledge sharing, research uptake, and collaboration among clinicians, researchers, and patient organisations. Eligible events must have a clear focus on rare disease or rare cancer research and meet both aims of the call: fostering knowledge exchange and increasing the inclusion of underrepresented countries.

more information

ERDERA 2026 Joint Transnational Call: Launching

On 10 December 2025, ERDERA will launch its 2026 Joint Transnational Call, "**Resolving unsolved cases in rare genetic and non-genetic diseases through variant validation and new technological approaches"**. The call will welcome proposals focused on providing diagnostic clarity in unsolved rare genetic and non-genetic diseases.

An information webinar for potential applicants will take place on 16 December 2025, 15:00–17:00 CET.

The full pre-announcement for the call is available on the ERDERA website. It describes the forthcoming opportunity for multinational research teams to apply, and will be updated with the full call when it launches.

website

Upcoming Meetings & Events

The High-Level Meeting on a European Research and Innovation Ecosystem for Rare Diseases

The High-Level Meeting (HLM) on a European research and innovation ecosystem for rare diseases, is going to take place on December 9-11, 2025, in Brussels, Belgium.

Secure your spot at the three-day High-Level Meeting on Rare Diseases by filling in the registration form. Please note registration is required for both in-person and remote attendance.

more information

Neurofibromatosis type 1 - Awareness day in Estonia

The conference on Neurofibromatosis Type 1 will be held at Tartu University Hospital, Tartu, Estonia, in a hybrid format (in-person and online) in English on December 19, from 10:00 to 15:30.

The event is organized in collaboration with ERN GENTURIS, ERN ITHACA, the Estonian Medical Genetics Society, and the Genetics and Personalized Medicine Clinic at Tartu University Hospital.

European Conference on Rare Diseases and Orphan Products (ECRD) 2026

SAVE THE DATE for the official conference days: 3–4 June 2026

more information

Visit our:
Website and LinkedIn

ERN GENTURIS

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