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ERN GENTURIS Newsletter

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

May 2025

ERN GENTURIS news

ERN GENTURIS Pre-ESHG 2025 Meeting

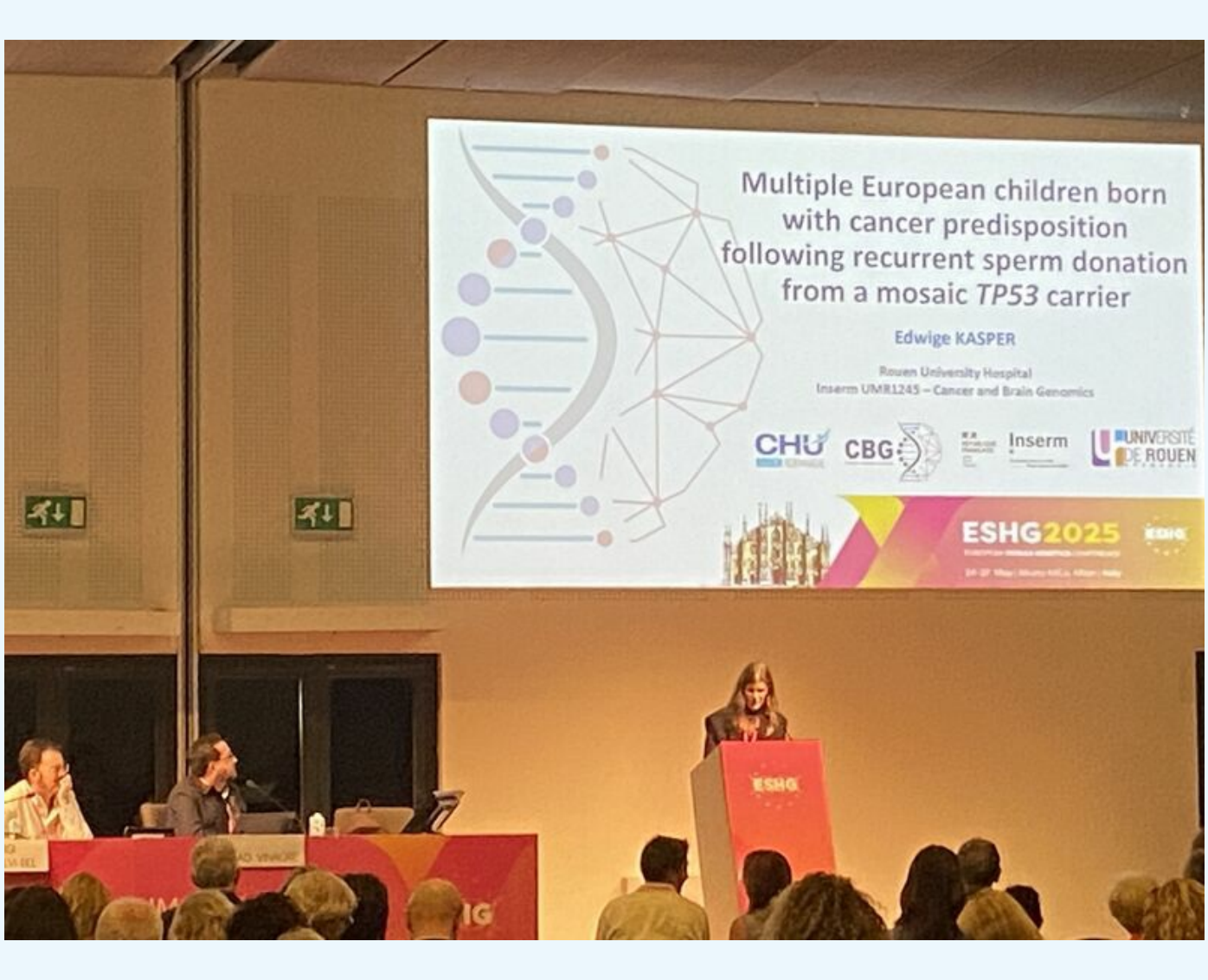
On 23 May 2025, ERN GENTURIS members came together in Milan for the pre-ESHG meeting, uniting to advance our shared mission of improving patient care for individuals affected by genetic tumor risk syndromes.

The meeting fostered dynamic discussions, collaboration, and strategic planning to enhance diagnosis, treatment, and support across Europe. Members reviewed ongoing projects, shared latest research insights, and explored new opportunities to strengthen cross-border cooperation.

During the meeting, our Registry leaders from Groningen, presented a detailed update on the GENTURIS registry and the Patient Portal, highlighting the progress made and the steps taken to strengthen data collection, patient involvement, and cross-border collaboration.

This gathering reinforced our commitment to putting patients first through innovation, education, and advocacy.

Sperm from cancer-risk donor used to conceive at least 67 children across Europe



Presented at ERN GENTURIS Pre-ESHG Meeting in Berlin Last Year – Now Featured at ESHG 2025!

We are proud to highlight a significant European collaboration addressing a critical and sensitive issue: multiple children across Europe born with cancer predisposition due to recurrent sperm donation from a mosaic TP53 variant carrier.

Initially presented at the ERN GENTURIS meeting in Berlin last year, this important case was recently showcased at ESHG 2025 (Session C13.06) by Edwige Kasper, demonstrating the impact of international cooperation in genomic medicine.

Key Insights:

- A single asymptomatic sperm donor, carrying a mosaic *TP53* variant linked to Li-Fraumeni Syndrome (LFS), unknowingly transmitted a cancer predisposition to multiple children born between 2008 and 2015 across Europe.
- All donations originated from the same private Danish sperm bank, which was unable to provide detailed donor information.
- **ERN GENTURIS** played a pivotal role in identifying the issue and coordinating responses across 5 countries involving 6 ERN members.
- Genetic counseling and reclassification efforts led to the *TP53* variant being classified as likely pathogenic.
- Among 58 children tested, 18 carry the variant and now benefit from tailored LFS surveillance; tragically, six have already been diagnosed with cancer.

This case underscores:
The urgent need for comprehensive genetic screening in gamete donation
Stronger regulation and improved communication between fertility clinics and sperm banks
The vital role of networks like ERN GENTURIS in enabling rapid, cross-border medical action

We extend our heartfelt thanks to all the centers, clinicians, and geneticists involved across Europe, including Svetlana Lagercrantz, Claude Houdayer, Robin de Putter, Kathleen Claes, Charlotte Kvist Lautrup, and Karin Wadt.

For more detailed coverage, read the article in The Guardian:

article

ERNs booth at ESHG 2025

ERN GENTURIS was proud to be featured alongside three other European Reference Networks (ERNs) (ERN EYE, ERN Euro-NMD and ERN ITHACA) at the European Society of Human Genetics (ESHG) 2025 conference in Milan. This prestigious event brought together leading experts in genetics and genomics to share the latest advancements and collaborative efforts across Europe.
Our presence highlighted ERN GENTURIS's commitment to improving care for patients with genetic tumor risk syndromes through cross-border collaboration, research, and patient advocacy.
We thank all members who contributed to making this a successful representation of our network's impact in genomic medicine.

In the spotlights - Darja Molan

Meet Darja Molan – New Chair of Patient Representatives at ERN GENTURIS.
We're proud to introduce Darja Molan, the new Chair of the Patient Representatives at ERN GENTURIS and President of Europa Donna Slovenija.
A BRCA1 mutation carrier and breast cancer survivor, Darja brings lived experience and strong advocacy to her new role. In our interview, she highlights the importance of genetic testing, psychological support, and cross-border collaboration to improve care for those with hereditary cancer risk.
Her message: prevention, awareness, and patient-centered care must be at the core of our efforts.

Read the full interview:

in the spotlights

ERN GENTURIS CPMS news

Thematic group	Number of patients discussed in ERN GENTURIS CPMS meetings in 2025
TG1: Schwannomatosis and neurofibromatosis	3
TG2: Lynch syndrome and polyposis	6
TG3: Hereditary breast and ovarian cancer	1
TG4: Other rare – predominantly malignant – genturis	5

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 **15** 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 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.

Education opportunities

ERN GENTURIS educational webinars

23 June 2025
Mosaicism in oncogenetics
Janneke Schuurs-Hoeijmakers

Watch the previous webinars here:

webinars

LEARN: MOOC Diagnosing Rare Diseases

Now open! A facilitation window for the MOOC Diagnosing Rare Diseases: from the Clinic to Research and back is open from Monday, May 12, until Friday, July 4.

During this period, experts and mentors will be available online to answer participants' questions and stimulate insightful discussions throughout the course.
This free online course, developed within the European Joint Programme on Rare Diseases (EJP RD), was co-created by ERN GENTURIS, ERN ITHACA, EURORDIS, and the Fondation Maladies Rares.
The MOOC explores key topics in the diagnosis of rare genetic diseases, including:

- The diagnostic process and types of genetic tests available
- Differences in patient pathways for rare genetic diseases
- Technological advances in diagnostic research
- The importance of collaborative studies and data sharing
- The impact of having (or lacking) a diagnosis on patients' lives
- The contribution of physiopathological and social science approaches in rare disease diagnostics

course

General news

European Commission Adopts Horizon Europe Work Programme 2025

On 14 May 2025, the European Commission officially adopted the Work Programme 2025 for Horizon Europe, setting the framework for research and innovation funding across the continent.

European Commission

Cluster 1: Health, features prominently, focusing on critical areas including:

- Staying healthy in a rapidly changing society
- Living and working in a health-promoting environment
- Tackling diseases and reducing disease burden
- Ensuring equal access to innovative, sustainable, and high-quality healthcare
- Developing and using new tools, technologies, and digital solutions for a healthy society
- Maintaining an innovative, sustainable, and competitive EU health industry

This programme offers significant opportunities for advancing health research and innovation aligned with patient needs and societal challenges.

For full details, check the European Commission's announcement here:

work programme

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**

Upcoming Meetings & Events

Visit our:
Website and LinkedIn

ERN GENTURIS

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