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ERN GENTURIS Newsletter

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

March 2025

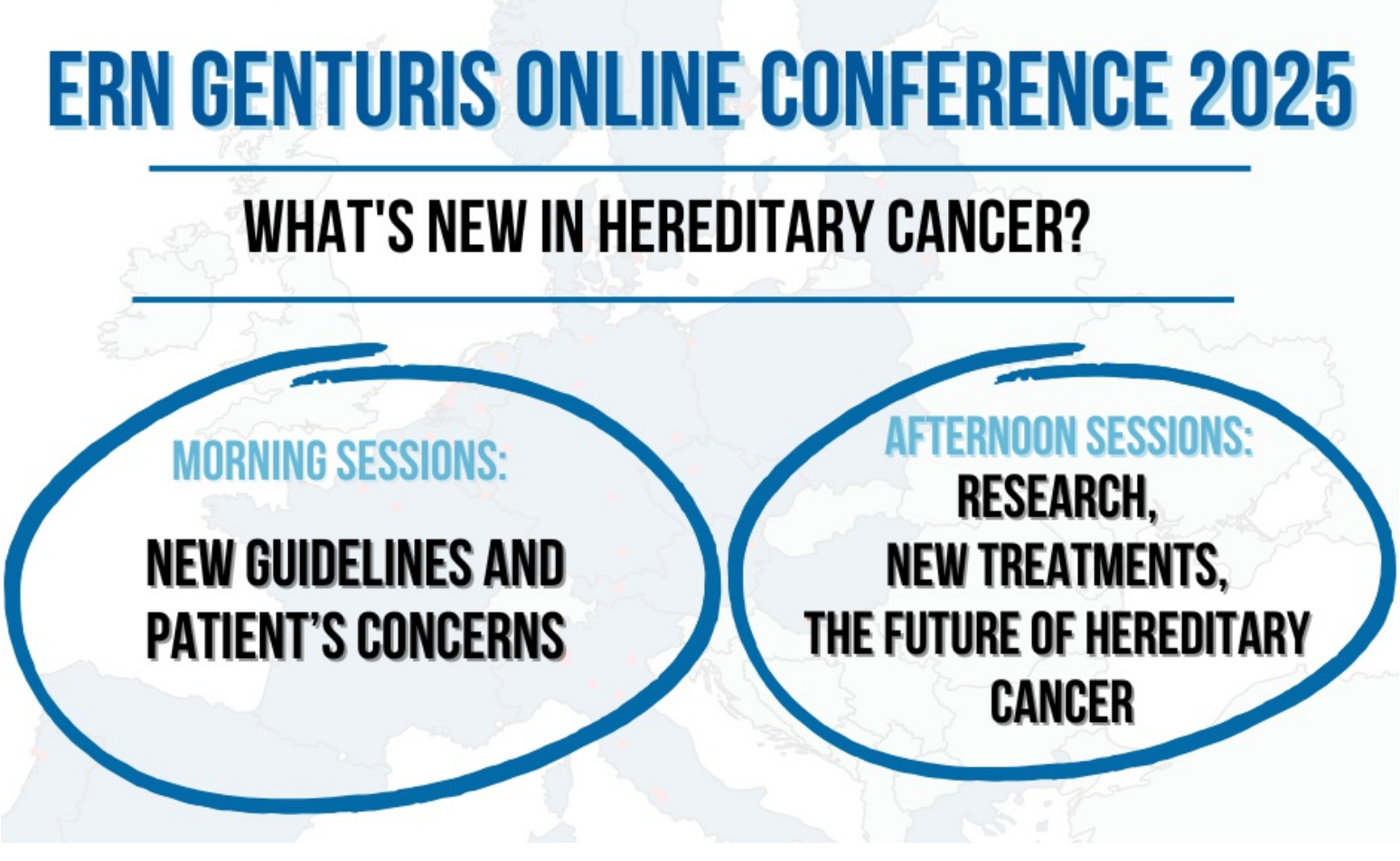
ERN GENTURIS news

ERN GENTURIS Conference 2025

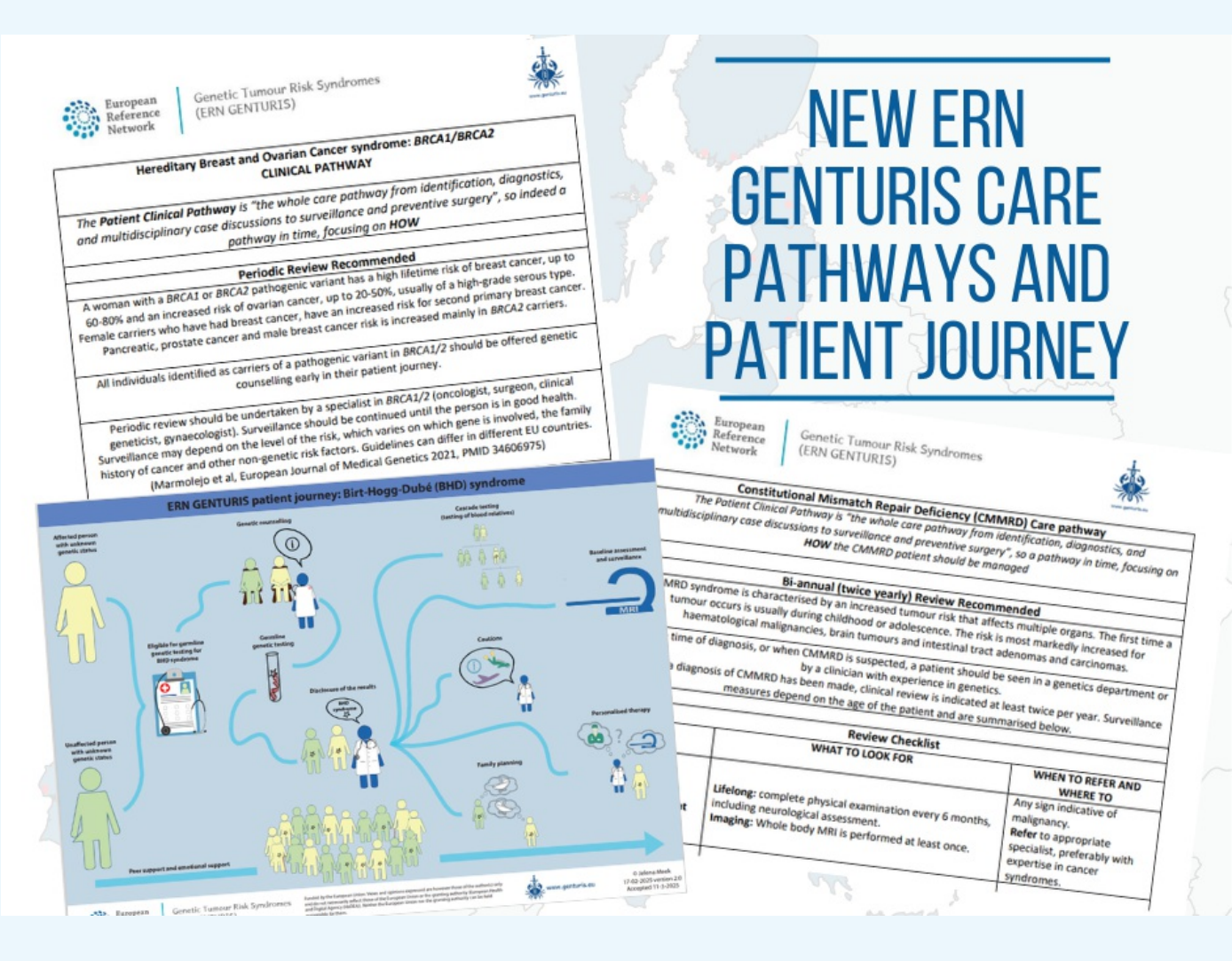
The ERN GENTURIS online conference 2025: What's new in hereditary cancer, was held on Thursday 20 March 2025 **and was a great success!**

Many great speakers were invited to the conference: Claas Röhl, Krajc Mateja, Chrystelle Colas, Marianne Geilswijk, Rianne Oostenbrink, Anke Oerlemans, Rolf Sijmons, Matt Bolz-Johnson, Richarda de Voer, Carla Oliveira, Jolanda de Vries, Lise Borgwardt, Verena Steinke-Lange, Karin Wadt, Svetlana Bajalica Lagercrantz, Marjolijn Ligtenberg, Judith Balmaña Gelpi, Antonella Cacchione, Eamonn Maher, Tiina Kahre, and Nicoline Hoogerbrugge.

Videos of the conference presentations will be made available when the editing of the recordings has been completed here.



ERN GENTURIS Care Pathways and Patient Journey



New ERN GENTURIS documents are now online:

1. **Hereditary Breast and Ovarian Cancer syndrome (HBOC)** care pathway
2. **Constitutional Mismatch Repair Deficiency (CMMRD)** care pathway
3. **Birt-Hogg-Dubé syndrome (BHD syndrome)** patient journey

A big thank you to all who have contributed to these ERN GENTURIS documents, specifically:

- Thematic Group 3 "Hereditary Breast and Ovarian Cancer syndrome" lead by Judith Balmaña.
- The ERN GENTURIS CMMRD guideline group: Chrystelle Colas, Léa Guerrini-Rousseau, Manon Suerink, Richard Gallon, Christian P. Kratz, Éloïse Ayuso, CMMRD Guideline Group, Laurence Brugières, Katharina Wimmer.
- The ERN GENTURIS BHD guideline group: Marianne Geilswijk, Maurizio Genuardi, Emma Woodward, Katie Nightingale, Jazzmin Huber, BHD syndrome Guideline Group, Eamonn Maher, Nataliya Di Donato, Joan Brunet.

We want to express our gratitude to all **patient representatives** who helped improved the patient journey.

news

In the Spotlights interviews

In our newest 'Spotlights,' you can read interviews with Thematic Group 1 leaders: **Rianne Oostenbrink**, paediatrician and **Said C. Farschtschi**, consultant neurologist.

You can learn about specific genturis case or family that had an impact on their clinical practice or how being a member of ERN GENTURIS help doctors in providing care to patients.

Read the full interview:

in the spotlights

ERN GENTURIS Publications

Please find recently published articles:

1. Pregnancy-related issues in rare and low-prevalence diseases: results of ERN transversal working group on pregnancy and family planning survey, Dina Zucchi, et al., link
2. Non-serous ovarian cancer in PTEN Hamartoma Tumor Syndrome: additional evidence for increased risk, Ane J. Schei-Andersen, et al., link
3. The impact of the new WHO Classification of renal cell carcinoma on the diagnosis of hereditary leiomyomatosis and renal cell carcinoma, Jan Degenhardt, et al., link

publications

ERN GENTURIS CPMS news

Thematic group	Number of patients discussed in ERN GENTURIS CPMS meetings in 2025
TG1: Schwannomatosis and neurofibromatosis	2
TG2: Lynch syndrome and polyposis	2
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 **10** 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:

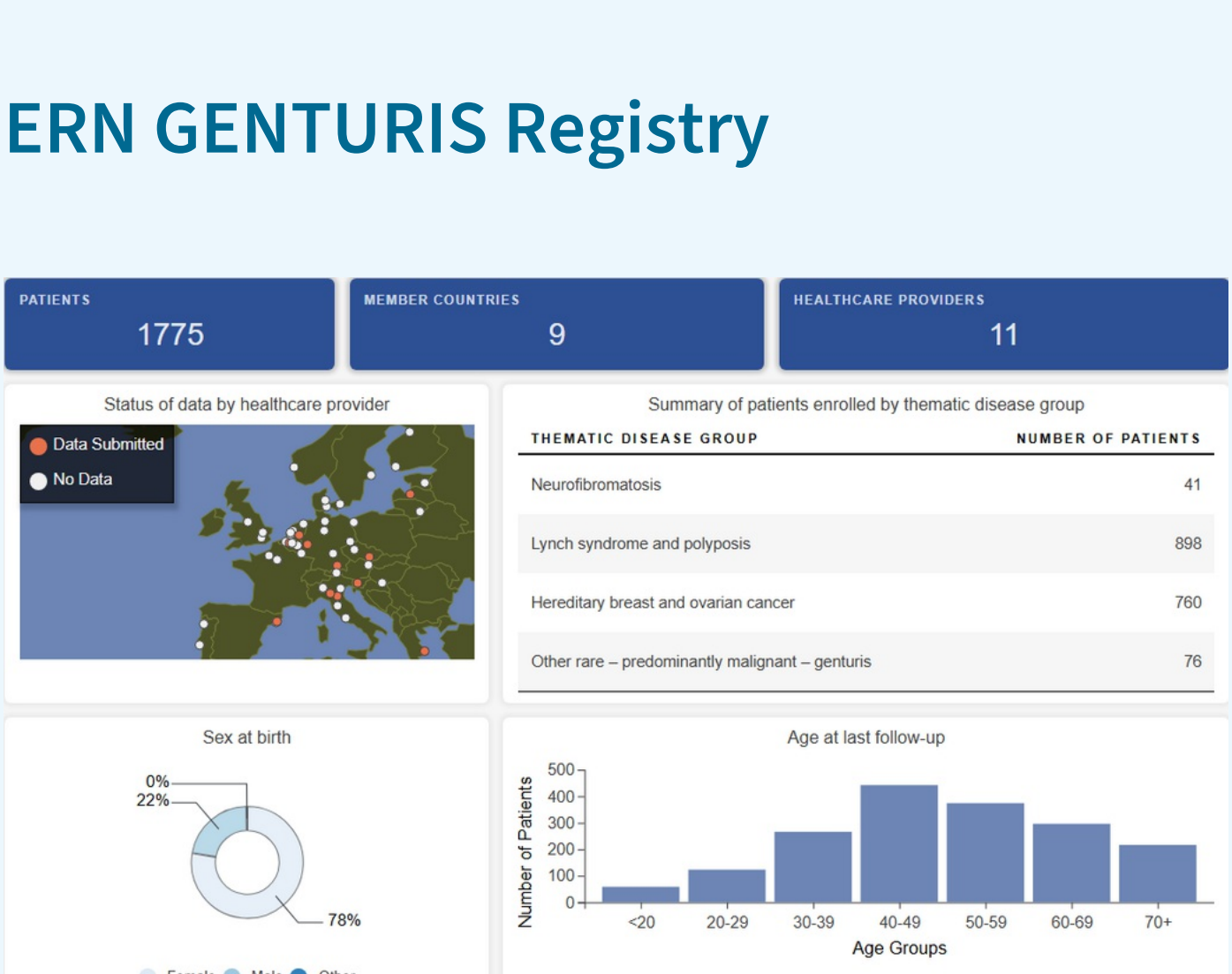
- How to refer a patient
- CPMS information and policy
- Guides on how to use CPMS 2.0

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.

ERN GENTURIS Registry



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 131 new patients have been included in the GENTURIS registry.

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

24 April 2025
Use of hormones in BRCA carriers: hormonal replacement therapy, oral contraception, ovarian stimulation – is it safe?
Sophie Frank

Watch the previous webinars here:

webinars

General news

SANT launches public consultation on rare diseases

European Parliament

European Parliament Committee on Public Health (SANT) launches public consultation on rare diseases.

The purpose of this public consultation is to provide a basis for the forthcoming work of the Committee on Public Health (SANT) in bringing a better understanding and detailed knowledge of the challenges of persons affected by rare diseases and views of persons working with or involved in rare diseases.

Fill in the survey by 31 March:

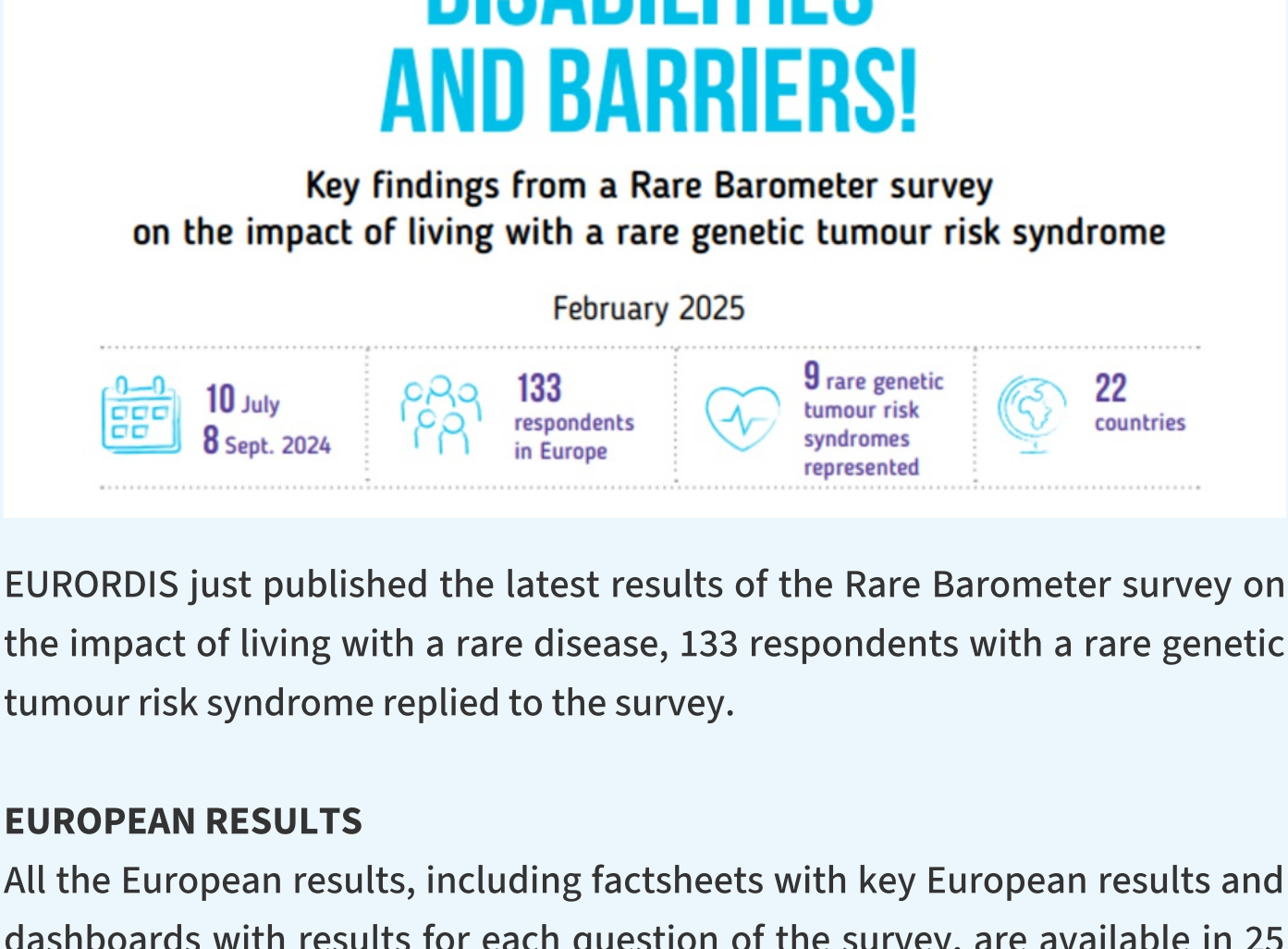
survey

Funding opportunities

European Commission Calls

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

Rare barometer results published



EURORDIS just published the latest results of the Rare Barometer survey on the impact of living with a rare disease, 133 respondents with a rare genetic tumour risk syndrome replied to the survey.

EUROPEAN RESULTS
All the European results, including factsheets with key European results and dashboards with results for each question of the survey, are available in 25 languages here: link
The full report in English will be published in March, at this link

results

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.

Register: [Conference on 'Towards an EU action plan on rare diseases' | EESC]

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**