Bekijk de webversie

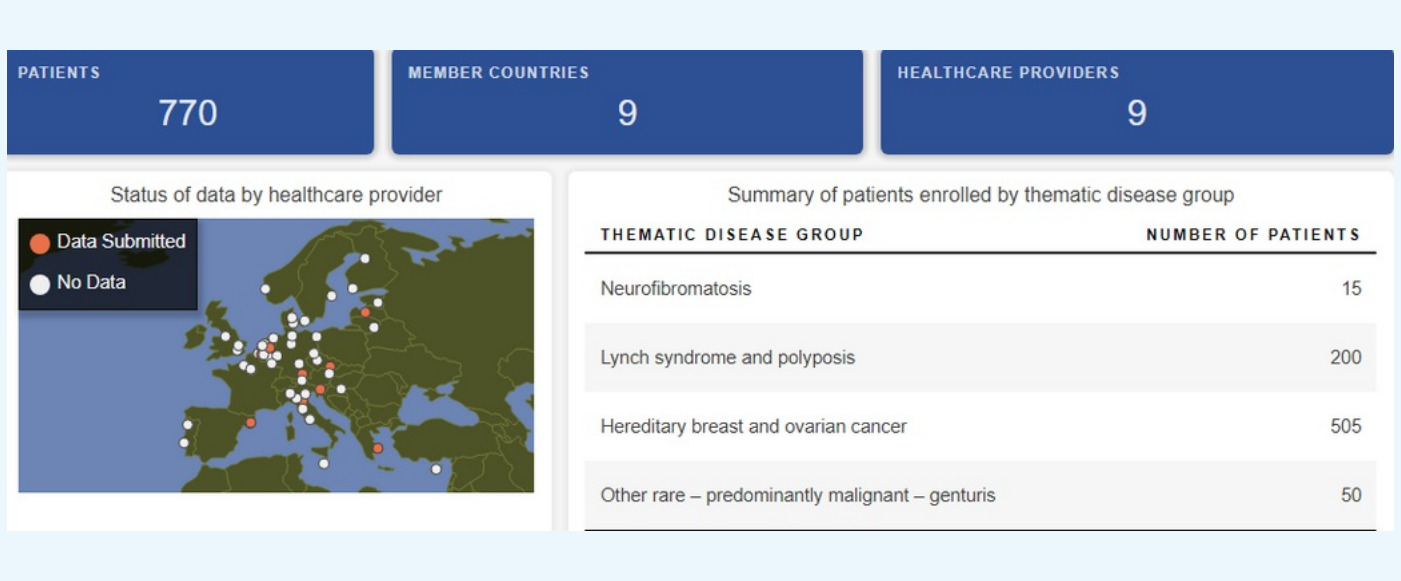
ERN GENTURIS Newsletter

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

April 2024

ERN GENTURIS news

ERN GENTURIS REGISTRY



The **GENTURIS registry** is the European registry for patients with one of the genetic tumour risk syndromes (genturis). The aim of the **GENTURIS registry** is to enable research that will provide insights into the natural disease history and care pathway by gathering data from a large number of genturis patients and providing regulated data access following a data access policy.

Since the 1st of November 2023 more than **400 new patients** have been included in the GENTURIS registry.

For more information, see https://genturis-registry.eu/ or contact the UMCG team via genturis-registry@umcg.nl

ERN GENTURIS overview of patients discussed in the CPMS in 2024

Thematic group	Number of patients discussed in ERN GENTURIS CPMS meetings in 2024
TG1: Schwannomatosis and neurofibromatosis	4
TG2: Lynch syndrome and polyposis	6
TG3: Hereditary breast and ovarian cancer	2
TG4: Other rare – predominantly malignant – genturis	9

In the discussions organized through the Clinical Patient Management System (CPMS), ERN GENTURIS has provided tailored expert advice to **21** genturis patients and their families in 2024 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
- CPMS guides

Education opportunities

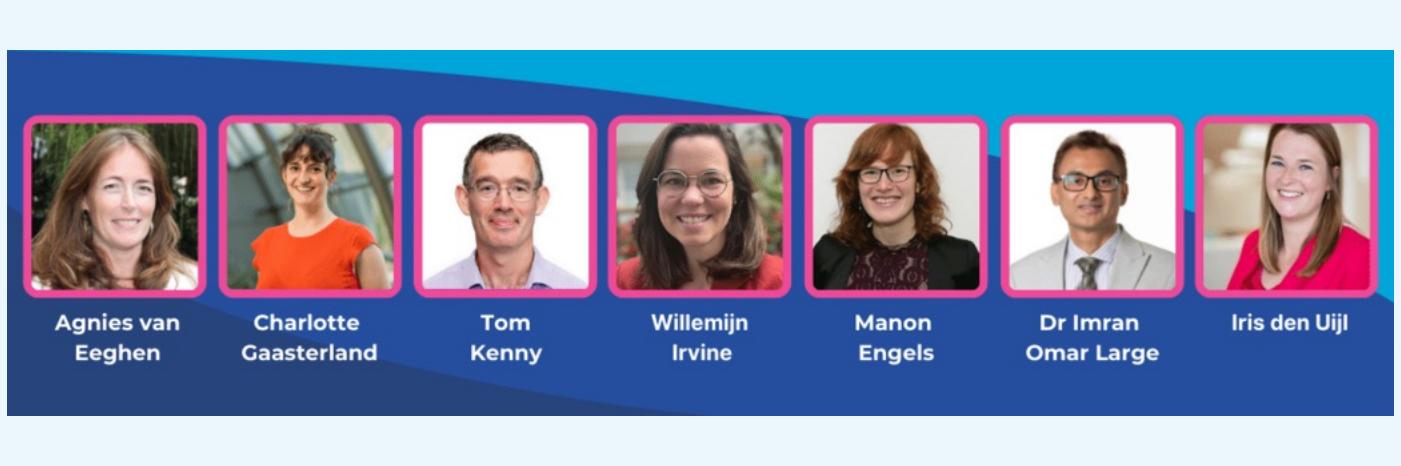
ERN GENTURIS educational webinars

22 May 2025
Molecular Diagnosis of the BRCA2 c.156_157insAlu, a Portuguese founder Pathogenic Variant
Fátima Vaz & Teresa Duarte

More on our website:

webinars

Approaches to Evaluate Evidence for Rare Diseases



EURORDIS-Rare Diseases Europe webinar: Approaches to the Evaluation of Evidence for Rare Disease and Complex Conditions Guidelines will take place on 29 April 2024 at 14.00-15.30 CET.

ERN GENTURIS Project Manager **dr Manon Engels** together with **Tom Kenny** (SquareRootThinking) will give a presentation about **ERN GENTURIS** experiences of the challenges in synthesis high quality evidence gathered in rare and complex conditions and approaches taken and potential solutions.

This webinar will bring together experts and methodologists experienced in successfully navigating the underlying issues. They will propose solutions for developing effective guidelines for the evaluation of evidence to support clinical decision making for people living with a rare or complex condition.

This webinar is open for the ePAGs, ERN Managers and guideline methodologist leads.

Read more here

registration

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

Important lessons on healthcare from rare diseases

A recently published article in the Lancet Neurology, explains the importance of **JARDIN** Joint Action, which is a pan-European initiative that involves 58 research and medical institutions in the 27 member states, Norway, and Ukraine. Its ultimate aim is the integration of European Reference Networks (ERNs) into each national healthcare system, to facilitate cross-border healthcare and speed up research advances.

article

In the context of enhancing the implementation of **Directive 2011/24/EU on patients' rights in cross-border healthcare (CBHC)**, under the EU4Health programme, the European Commission with assistance of the National Contact Points, is customising and improving written communication materials and organising a series of **10 workshops at national level, culminating in an EU-level event in 2025**.

The materials and workshops focus on **patients' rights in cross-border healthcare, rare diseases, and European Reference Networks (ERNs).**

The first workshop took place in Malta, read the flash report.

flash report

Data on cross-border patient healthcare following Directive 2011/24/EU for reference year 2022

The cross-border health care directive (Directive 2011/24/EC) requires the Commission to periodically report on the operation of the Directive to the European Parliament and to the Council.
The recently published report confirms the directive is still "relevant and fit-for-purpose" but there are still barriers to access across borders.
The 24 European Reference Networks are a "major achievement" for cross-border health care cooperation, according to the report. However their integration into national systems is necessary to safeguard their sustainability and to fully benefit rare disease patients.

report

Targeted Consultation on EU4Health: have your say on future priorities, orientations and needs

The European Commission invites interested stakeholders to share their views on future priorities, orientations and needs for the future EU4Health programme by 10th June 2024. This includes Member State authorities responsible for health at national, regional or local level.

The feedback collected will not only contribute to the consultation report but will also feature in discussions at the EU4Health Stakeholder's Conference on 19th June 2024.

Share your insights between **22nd April and 10th June 2024.**

link

On 24 April 2024 the European Parliament adopted **the European Health Data Space (EHDS) regulation**, marking a historical moment for digital health in the EU.
The EHDS will empower individuals to access their electronic health data through patient portals or apps. This is aligned with the goals of the Digital Decade policy programme 2030: 100 % citizens having access to their electronic health records. Furthermore, the regulation will ensure that electronic health data follows patients when they seek care from different healthcare providers within their own Member State or across the EU.

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

The European Conference on Rare Diseases & Orphan Products

The ECRD is the largest, patient-led, rare disease policy-shaping event held in Europe, bringing together people with rare diseases and patient advocates, policymakers, healthcare industry representatives, clinicians, regulators and Member State representatives. The conference is taking place on **15 & 16 May 2024.**

Registration is still open for the online attendance.

register

Conference of Polish ERN members

The conference 'Rare Diseases - problems not only systemic' is taking place in Cracow, Poland on **17 & 18 May 2024.**

The event is organised by the Polish ERN members. It will be attended by outstanding scientists, specialists in specific fields of medicine, as well as decision-makers shaping the health care system in Poland, representatives of patient organizations and representatives of key organizations in the health sector.
ERN GENTURIS Project Manager, **M.sc., M.sc. Katarzyna Urbanczyk**, will present the results of her recent study about the experience of the Polish ERNs. Guest speakers will present the approach to rare diseases in Spain (**dr Enrique Terol, MD**) and the Netherlands (**prof. dr. Wendy van Zelst-Stams**).

more information

The EJP RD Final Conference

The conference is set to bring together leading experts, professionals, and enthusiasts from around the world. Find more about the event here.

Join on **May 27-28, 2024** for engaging sessions on rare disease research.
Check out the **agenda** for a sneak peek into the sessions and plan your conference experience.

The registration deadline for the Final EJP RD Conference has been extended to **May 22nd, 2024**

register

European Human Genetics Conference

The next ERN GENTURIS meeting will take place prior to the ESHG conference in Berlin on 31 May and 1 June 2024.

On Sunday, 2nd June at 12:15 - 13:15 CEST the Get2gether European Reference Networks session will take place in Room A1.

Stop by the ERN booth (1-3 June) shared by ERN GENTURIS, ERN ITHACA, ERN-EYE and EURO-NMD, you will find us in the hall 4.2, with the posters.

registration

5th course in Hereditary Cancer Genetics

This course, organized by members of **ERN GENTURIS**, aims at delivering up-to-date knowledge on hereditary

cancers to clinical and molecular geneticists in training or certified. It creates the best opportunity for interaction and discussion with experts from all over Europe, in the fabulous environment of Bertinoro, the headquarters of the ESHG sponsored courses. The faculty combines experts in clinical and molecular genetics, known for their didactic skills. Participants are encouraged to present a clinical or genetic case in a Poster format for on-site discussion.
The course will take place on 17-20 September 2024.

more information

Visit our:
Website and LinkedIn

ERN GENTURIS
With every diagnosis we can help an entire family