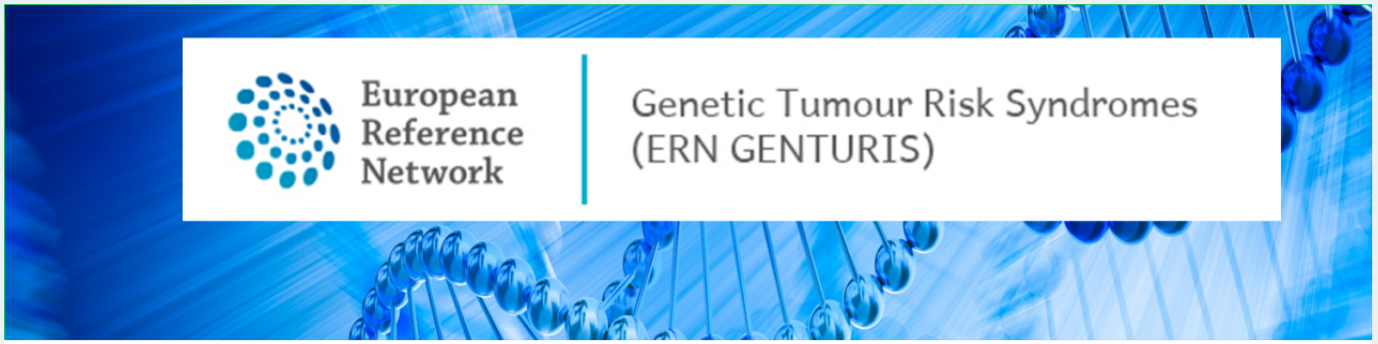


# ERN GENTURIS Newsletter May 2018

## About ERN GENTURIS

Genetic tumour risk syndromes (genturis) are disorders in which inherited genetic mutations strongly predispose individuals to the development of tumours. The lifetime risk of cancer can be as high as 100 %. While there is considerable diversity in the organ systems that may be affected, individuals affected by these conditions share similar challenges: delay in diagnosis, lack of prevention for patients and healthy relatives, and therapeutic mismanagement.

The European Reference Network GENTURIS is working to improve identification of these syndromes, minimise variation in clinical outcomes, design and implement guidelines, develop registries and biobanks, support research, and empower patients.

## GENTURIS website available in multiple languages

We are proud to announce that our website is now available in a French and Spanish version, and will soon be available in German, Portuguese, Polish and Slovenian.

**Visit website**

## Patient participation - ePAG representatives

ERN GENTURIS is committed to a broad patient engagement and a full partnership approach between patients and professionals. The members of the ERN GENTURIS European Patient Advocacy Group (ePAG) participate on all levels in GENTURIS decision-making processes to ensure that the patient voice is heard throughout the ERN GENTURIS development process.

A new European Patient Advocacy Group Representative has joined the GENTURIS ePAG! Nicola Reents from the German Familienhilfe Darmkrebs e.V./ semi-colon will join ePAG representative Jurgen Seppen to represent Thematic Group 2: Lynch syndrome and polyposis.

**See ePAG members**

## Genturis scientific literature - monthly overview

ERN GENTURIS member Thierry Frebourg provides a monthly selection of newly published papers about genetic tumor risk syndromes, available through our website.

**Selection of papers**

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## National networks

The current list of participating healthcare providers covers 12 member states. An important aim for the second year of operation is to develop national networks in these members states. The national coordinators are currently working to identify all relevant stakeholders in their countries while on GENTURIS level, the dissemination and communication methods and channels will be further expanded to facilitate a 2-way communication with the stakeholders.

List of Health Care Providers

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## Spring Course in Hereditary Cancer Genetics

For the second time, 25 GENTURIS experts offered a Spring Course in Hereditary Cancer Genetics from 24-27 April 2018 in the beautiful setting of Bertinoro, Italy.

With 85 participants originating from 20 European countries and 12 countries outside Europe, the course was fully booked and was evaluated very positively

The Spring course is organized every 2 years, is aimed at clinical and molecular geneticists in training or certified, and covers Basics in cancer genetics, Breast/ovarian cancer and rare genetic tumor risk syndromes, Gastrointestinal cancer and panel testing, Prevention and treatment, and offers interactive workshops.

Program

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## Stakeholders meeting in Lisbon, Portugal on May 11th, 2018

A dedicated workshop to discuss the impact of the ERNs in Portugal was held on 11 May 2018 during the biannual meeting of the Portuguese Association of Cancer Research. The workshop was organised by Carla Oliveira (National Coordinator Portugal for ERN GENTURIS) and Tamara Hussong Milagre (Patients' Association EVITA, FADOP and ePAG Representative ERN GENTURIS). ERN GENTURIS coordinator Nicoline Hoogerbrugge introduced ERN GENTURIS to the audience. About 100 people attended the workshop; patients and representatives from patient associations as well as doctors, nurses, and company employees.

*Tamara Hussong Milagre, Nicoline Hoogerbrugge and Carla Oliveira*

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## Workshop on diagnostic criteria for Neurofibromatosis

Neurofibromatosis (NF) experts Prof Gareth Evans, Prof Eric Legius and Prof Pierre Wolkenstein, are leading on the development of revised diagnostic criteria for NF1, NF2 and schwannomatosis. These GENTURIS experts are part of the leadership group of European and American experts that are leading a Delphi process and organizing a meeting in New York City on June 11-12, 2018 to re-evaluate the existing criteria that date from 1987 (NF1), 1992 (NF2) and 2011 (Schwannomatosis). The new consensus diagnostic criteria will be finalized at the International neurofibromatosis conference in Paris in November 2018 hosted by Pierre Wolkenstein.

# Solve-RD - Solving the unsolved Rare Diseases

A large consortium led by the University of Tübingen, the Radboud university medical center Nijmegen and the University of Leicester has acquired a € 15 million grant for the Solve-RD research program. SOLVE-RD aims to improve the diagnosis and treatment of rare diseases, which in total affect millions of Europeans.

Solve-RD comprises four ERNs for rare neurological diseases (ERN-RND), neuromuscular diseases (ERN-EURO-NMD), congenital malformations and intellectual disability (ERN-ITHACA) and genetic tumor risk syndromes (ERN-GENTURIS).

ERN GENTURIS will focus on genetically unsolved, but most likely hereditary colorectal cancer, polyposis, gastric cancer and pheochromocytoma.


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