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Newsletter

European Reference Network

Genetic Tumour Risk Syndromes (ERN GENTURIS)

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Two new ERN GENTURIS guidelines

Surveillance guidelines for Heritable TP53-related cancer Syndrome (hTP53rc) and PTEN hamartoma tumour syndrome (PHTS) were developed over the previous 12 months, each by a dedicated core writing group. These guidelines piloted the process defined in the ERN GENTURIS Policy for Guideline Development: defining the guideline scope, completing a literature review and grading the evidence. The guideline and their recommendations were drafted with patient involvement and external review and consultation from a selected multidisciplinary group of experts in the Guideline Development Group. Both guidelines were assessed using the AGREE II criteria and scored highly. Finally, a plain language summary was completed for both guidelines. The guidelines will be released in the beginning of 2020.

Website cancergenetics is now available in 11 languages

The website www.cancergenetics.eu by ERN GENTURIS is now available in multiple languages! The website contains information for health professionals and patients who are considering a genetic test to look for an inherited cause of cancer.

Affiliated partner news

Three HCPs in Denmark (Aarhus Universitetshospital, Odense Universitetshospital and Rigshospitalet Copenhagen) and 1 consortium consisting of 3 HCPs from Cyprus (Archbishop Makarios III Hospital, Karaiskakio Foundation, Cyprus Institute of Neurology and Genetics) were designated as Associated National Centres to ERN GENTURIS. Luxemburg has designated Centre Hospitalier du Luxembourg as a National Coordination Hub for ERNs. On the basis of this designation, the HCP will be establishing a formal link with each of the 24 existing ERNs.
ERN GENTURIS was already designated the Mater dei Hospital in Malta as National Coordination Hub and 5 Associated National Centres,: 2 in Austria, 1 in Estonia, 1 in Latvia, and 1 in Norway.

Patient position paper

During the ERN GENTURIS annual meeting in Amsterdam on September 27th, the ePAG Patient Representatives Claas Röhl, Tamara Hussong Milagre, Nicola Reents and Rita Magenheim presented the patient position paper which they developed over the past 12 months.

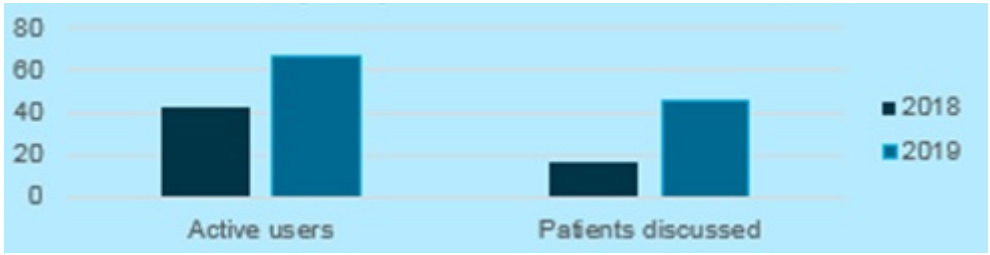
The Network Members welcomed the position paper as it gave a clear set of priority areas that the Thematic Groups and Executive Committee can focus on. In addition, the challenge from the patient representatives to think holistically at a Network level and not just at a HCP Member level was well received.

Open access paper on Patient Journeys online

ERN GENTURIS has developed an innovative visual approach for patient input into the Network, to share their expertise and experience: “Patient Journeys”. Our open access paper “Patient Journeys”: improving care by patient involvement" in the European Journal of Human Genetics is now online. It can be downloaded from https://doi.org/10.1038/s41431-019-0555-6.

CPMS

The use of CPMS keeps growing: in 2019 there were 66 active users and 46 patients were discussed.



Recurring meetings

CPMS recurring meetings are scheduled on the **first Friday of every month at 13.30-14.30**.

There are 2 parallel sessions, 1 for NF patient cases and 1 for patient cases related to the other thematic groups (Lynch Syndrome and Polyposis, Hereditary Breast and Ovarian Cancer, and Ultra-rare genturis).

Solve-RD: ultra-rare GENTURIS cases

"Solve-RD - solving the unsolved rare diseases" is a research project funded by the European Commission for five years (2018-2022).

The main ambitions of the project are to solve large numbers of rare diseases, for which a molecular cause is not known yet by sophisticated combined omics approaches, and to improve the diagnostics of rare disease patients through contribution to, participation in and implementation of a "genetic knowledge web" which is based on shared knowledge about genes, genomic variants and phenotypes. ERN-GENTURIS is one of the main ERNs who operates within Solve-RD.
One aim of Solve-RD is to solve ultra-rare diseases. At the annual Solve-RD meeting in February 2019 clinicians and researchers involved in ERN-GENTURIS defined the following phenotypes as 'ultra-rare GENTURIS cases':

1. individuals who developed primary cancers in at least four different organs or in three different organs when two cancers are rare
2. individuals who were diagnosed with any adult type of cancer below the age of 25 years
3. individuals who developed at least 100 adenomatous polyps before the age of 50 years

Within ERN-GENTURIS all centres can contribute in submitting samples for this cohort if sufficient germline DNA of the patient is available (3µg). When a sample is selected by the ERN-GENTURIS clinical board it will undergo whole-genome sequencing. The whole-genome sequenced data will be analysed by three ERN-GENTURIS dedicated Solve-RD PhD students. In case a pathogenic causative variant is identified in a sample this will be communicated back to the submitter. The submission of 'ultra-rare GENTURIS cases' is open and enquiries or questions can be directly addressed to richarda.devoer@radboudumc.nl.

Teaching

7th up-to-date in Hereditary cancer
A course for Dutch care professionals on hereditary cancer will be given on 24 January 2020 at the Radboudumc, Nijmegen, the Netherlands. Click here for more information.

Bertinoro
The ESHG-ERN GENTURIS Bertinoro course for clinical and molecular geneticists (and those in training) will be given on 21 – 25 September 2020 in Bertinoro, Italy. Click here for more information.

3rd annual ERN GENTURIS meeting in Amsterdam

27 and 28 September 2019

The third annual ERN GENTURIS meeting took place in the Trippenhuis in Amsterdam centre on 27 and 28 September. The draft guidelines for Heritable TP53-related cancer Syndrome (hTP53rc) and PTEN hamartoma tumour syndrome (PHTS) were shown, and the patient representatives presented their position paper.

Upcoming meetings:

- **ECRD 2020 in Stockholm**

ERN GENTURIS is an associate partner of the 10thEuropean Conference on Rare Diseases & Orphan Products, which will be held in Stockholm, Sweden on **15 and 16 May 2020**. The overarching theme is the rare disease patient journey in 2030.

Deadline for poster submissions is 10 February 2020.

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ERN GENTURIS Coordinating Center:
Radboud university medical center
The Netherlands

genturis@radboudumc.nl
https://www.genturis.eu

EURORDIS-Rare Diseases Europe is also offering patient fellowships for up to 40 patients' advocates to attend this conference. These fellowships aim at empowering patient advocates by offering a platform for networking opportunities, access to information and sharing experiences.

- **European NF meeting 2020 in Rotterdam**

The 19th European neurofibromatosis meeting will take place in Rotterdam on 10-13 September 2020, organised by ERN GENTURIS Member Erasmus MC national NF1 Expertise centre (ENCORE) and the patient organisation for neurofibromatosis in the Netherlands (NFVN). For more info, see https://nf2020.nl/

- **ERN GENTURIS - Closed meeting Berlin**

The ERN GENTURIS closed meeting preceding ESHG Berlin will take place on 5-6 June 2020.

- **ERN GENTURIS - Annual meeting Amsterdam**

The ERN GENTURIS annual meeting 2020 will take place on 8-9 October 2020.