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

Newsletter September 2022

Webinar: The process of variant classification and its implications for clinical management

Wednesday 5 Oct 2022, 16:00-17:00 CEST

Prof. dr. Maurizio Genuardi
Policlinico Gemelli University Hospital, Rome, Italy

Maurizio Genuardi is an academic clinical geneticist, full professor of Medical Genetics at the Catholic University in Rome and head of Medical Genetic Services at the Policlinico Gemelli University Hospital in Rome, Italy. He is currently Vice-President of the European Society of Human Genetics.

The expanding use of genomic tests has led to an exponential increase in the number of identified DNA sequence variants. This webinar will deal with: 1. the datasets used for variant classification according to their pathogenicity; 2. General vs gene-specific criteria for interpretation; 3. Clinical utility and actionability of the genes tested (and of the variants identified): this is especially important since several variants are identified nowadays in genes whose clinical consequences are disputed or not well defined.

If you would like to attend this webinar, please register here:
https://register.gotowebinar.com/register/4059911925102251275

Webinar: Prophylactic mastectomies – the surgeon's perspective

Wednesday 26 Oct 2022, 16:00-17:00 CEST

Prof. dr. Janez Žgajnar
Institute of Oncology Ljubljana, Slovenia

Prof. Janez Žgajnar, MD. PhD is a breast surgeon from the Institute of Oncology Ljubljana, Slovenia. He is a member of the Executive committee of the Breast surgery Division of the UEMS, vice president of the Slovenian National cancer control plan, president of the Slovenian cancer society and vice president of the Slovenian Senology society.

After discovery of hereditary breast cancer related genes, several preventive strategies are in use in affected individuals and families. Prophylactic surgery is to date the most effective breast cancer risk reducing intervention, which may reduce the risk by at least 90%. There are several technical issues to be considered related to the prophylactic surgery. Furthermore, the surgeon is involved in psychological aspects along with the counselling and treatment process of the affected individuals and families.

If you would like to attend this webinar, please register here:
https://register.gotowebinar.com/register/9052244471124366862

Upcoming webinars

09-11-2022
Angelique Flöter Rådestad
Hereditary breast- and ovarian cancer, aspects on risk reducing measures and hormonal replacement – the gyne-oncologist's perspective

23-11-2022
Ann Nordgren & Svetlana Bajalica Lagercrantz
Childhood cancer and genetic tumour risk syndromes – the geneticist's perspective

06-12-2022
Chella van der Post
Hereditary gastrointestinal cancer – the pathologist's perspective

For more information and registration, visit www.genturis.eu

European Reference Network
for rare or low prevalence complex diseases
Network Genetic Tumour Risk Syndromes (ERN GENTURIS)

Von Hippel-Lindau disease: Updated guideline for diagnosis and surveillance

endorsed by ERN GENTURIS

The ERN GENTURIS Special Issue of the European Journal of Medical Genetics now contains 11 papers by ERN GENTURIS members. One of the latest publications in the special issue is the updated guideline on Von Hippel-Lindau disease by Marie Louise M Binderup *et al*. This guideline is based on evidence from the international vHL literature and extensive research of geno- and phenotypic characteristics, disease progression and surveillance effect in the national Danish vHL cohort. doi: 10.1016/j.ejmg.2022.104538.

Annual ERN GENTURIS meeting Amsterdam, 8-9 September 2022

After 2 years of having the annual network meeting online, ERN GENTURIS HCP representatives and patient representatives were happy to meet face-to face in Amsterdam to discuss network updates and activities.

ERN GENTURIS Grant signed for period March 2022 - August 2023

On 23 August, the so-called “bridging grant” for ERN GENTURIS was signed by Radboudumc and the European Commission. ERN GENTURIS was among the first of the 24 ERNs to secure this funding. The grant will finance management of ERN GENTURIS for the period 1 March 2022 to 31 August 2023. Radboudumc has prefinanced the salaries of the coordination team during the first 6 months of this period. It is expected that in 2023 there will be a call for the management grant for the subsequent 3.5 year period .

Besides the ERN Research Mobility Fellowship Funding opportunity, the ERN Research Training Workshops funding opportunity is still open for applications until October 1st. The goal of the workshops is to train researchers and clinicians affiliated to ERN- Full Members or – Affiliated Partners in relevant topics on research in rare diseases.