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Newsletter

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

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ERN GENTURIS - upcoming webinars and endorsed guidelines

In this ERN GENTURIS newsletter you will find two updates:

- Open access paper on Germline TP53 Testing in Breast Cancers
- Upcoming webinars

Open access paper on Germline TP53 Testing in Breast Cancers

The open access review paper "*Germline TP53 Testing in Breast Cancers: Why, When and How*?" by ERN GENTURIS members Gareth Evans, Emma Woodward, Svetlana Bajalica-Lagercrantz, Carla Oliveira and Thierry Frebourg has been published in Cancers as part of the Special Issue *Genetic Variants Associated with Breast and Ovarian Cancer Risk* and is available online: https://doi.org/10.3390/cancers12123762

Webinar 'Gastrointestinal Polyposis Syndromes'

13 January – 16:00 CET
Prof. Stefan Aretz, Bonn

Stefan Aretz is professor of Genetics of Familial Tumour Syndromes at the University of Bonn and will be the speaker of this webinar on gastrointestinal polyposis syndromes.

Several clinically characterised hereditary tumour predisposition syndromes with predominant manifestation in the gastrointestinal tract have been delineated within the last decades and explained by germline mutations in specific tumour suppressor or DNA repair genes. While some syndromes are restricted to gastrointestinal neoplasms, others show a variety of extraintestinal lesions. A clinical diagnosis might be challenging in the absence of a distinct family history and in case of a mono-symptomatic course. However, multigene panel testing can uncover those cases. For most of the polyposis syndromes, well established and efficient surveillance programs are available while in others, consensus recommendations are still lacking. This review will address state-of-the-art diagnostic algorithms and discuss present surveillance and management strategies.

If you would like to attend this webinar, please, register here.

Webinar 'Variants identified in tumors can be germline - when to refer for germline testing'

27 January – 16:00 CET
Dr. Kathleen Claes, Ghent

Dr. Kathleen Claes is affiliated to the Center for Medical Genetics of Ghent University Hospital, Belgium. She is a European registered Clinical Laboratory Geneticist, with a main expertise in molecular genetics of cancer predisposition syndromes.

Furthermore, she was involved in the implementation of the molecular diagnostics platform for tumor sequencing in her hospital. Since the number of druggable tumour-specific molecular aberrations has grown substantially in the past decade, tumour molecular profiling has been introduced at a large scale and revolutionized the field of precision oncology. Besides variants driving therapeutic decisions, germline variants are being detected, with implications for both the patient and relatives. During the webinar an overview will be provided of the most relevant

genes included in many cancer gene panels. Additionally, the prevalence of germline variants in different tumour types and issues on clonal hematopoiesis and mosaicism will be discussed and case-illustrated. This webinar aims to increase knowledge on the complexity of variant interpretation and to show that tumour sequencing must be accompanied by a plan for return of germline results, in partnership with genetic counselling.

If you would like to attend this webinar, register here.

Upcoming webinars:

13 January: Gastrointestinal polyposis syndromes – *Prof. Stefan Aretz*.

27 January: Variants identified in tumours can be germline – when to refer for germline testing – *Prof. Kathleen Claes*

10 February: Germline mutations as a therapeutic target in breast cancer - *Dr. Judith Balmaña*

24 February: Cancer Genetic Counseling and previvorship in an Era of Rapid Change – *Ass. Prof. Ignacio Blanco*

17 March: Von Hippel-Lindau Syndrome – *Prof. Eamonn Maher*

31 March: Psycho-oncologic aspects of genetic tumour risk syndromes – *Prof. Yvonne Brandberg*

For more information on these webinars or for registration, please visit the ERN GENTURIS website.

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