Newsletter December 2022

Webinar: Precision Oncology – conclusions for human genetics and genetic tumour risk syndromes

Wednesday 14 Dec 2022, 16:00-17:00 CET

Prof. dr. Evelin Schröck
Hereditary Cancer Syndrome Center Dresden, Germany

Evelin Schröck is a senior academic clinical geneticist, full professor of clinical genetics at the University Hospital Carl Gustav Carus at the Technische Universität Dresden, Germany. Her clinical and research work focusses on advancing genetic technologies and tumour genetics and is aimed at improving highly specialized healthcare for patients and families with hereditary cancer as well as intellectual disability.

Hereditary monogenetic variants can lead to an increased cancer risk for all affected family members and have implications for cancer surveillance, treatment options and genetic testing. In this webinar, Evelin Schröck will summarize the results of a large-scale precision oncology study performed within the German DKTK MASTER program. In this observational trial prospective and comprehensive massive parallel tumour and control exome/genome sequencing is performed and is used for the recommendation of targeted treatment options. Germline variant evaluation is part of the workflow and a retrospective analysis of germline variant evaluation and its clinical impact for nearly 1,500 retrospectively analyzed patients will be presented.

Evelin Schröck will emphasize on the relevance of precision oncology studies to identify patients at risk for genetic tumour risk syndromes and their contribution to an improved multidisciplinary clinical decision making for these patients.

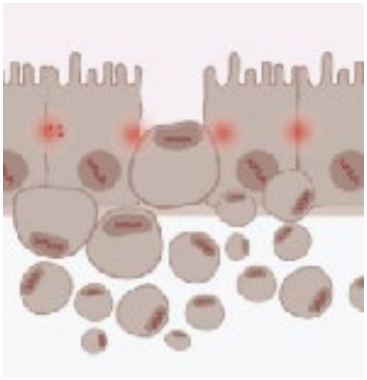
If you would like to attend this webinar, please register here:
https://attendee.gotowebinar.com/register/4872215725790632206

Upcoming webinars

25-01-2023
Stefan Aretz
Genetics 30 years after the discovery of APC

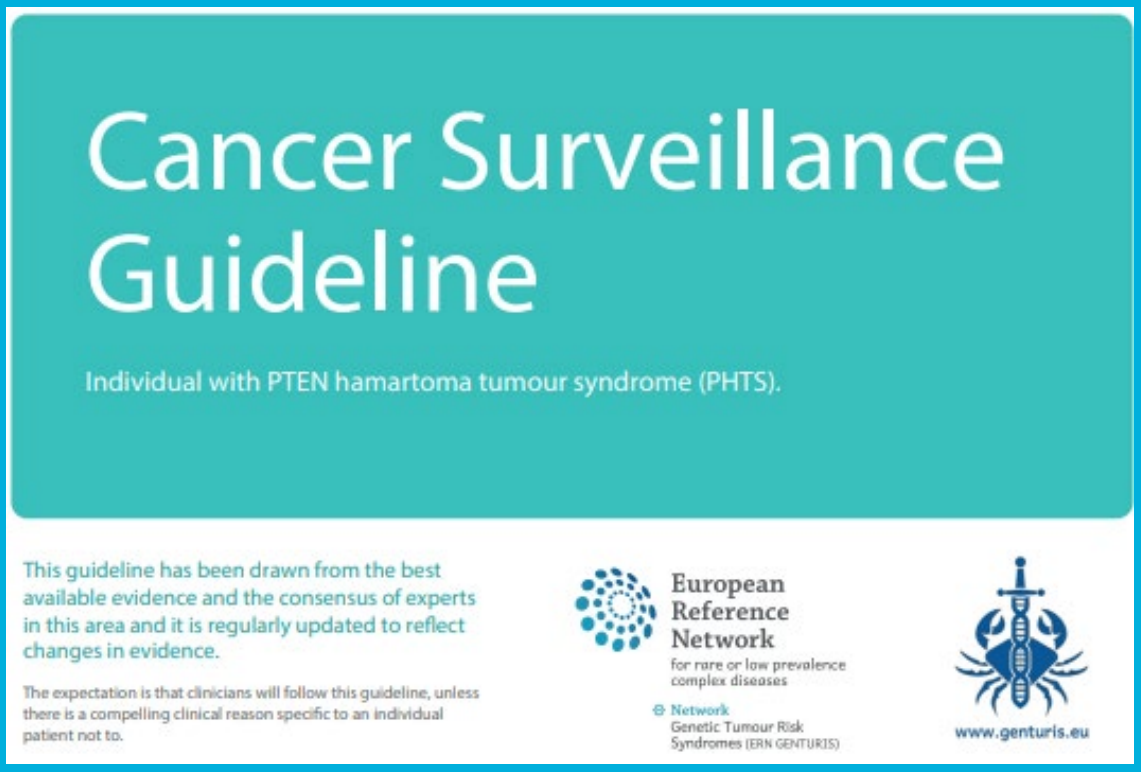
06-2-2023
Chella van der Post
Hereditary gastrointestinal cancer – the pathologist's perspective

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



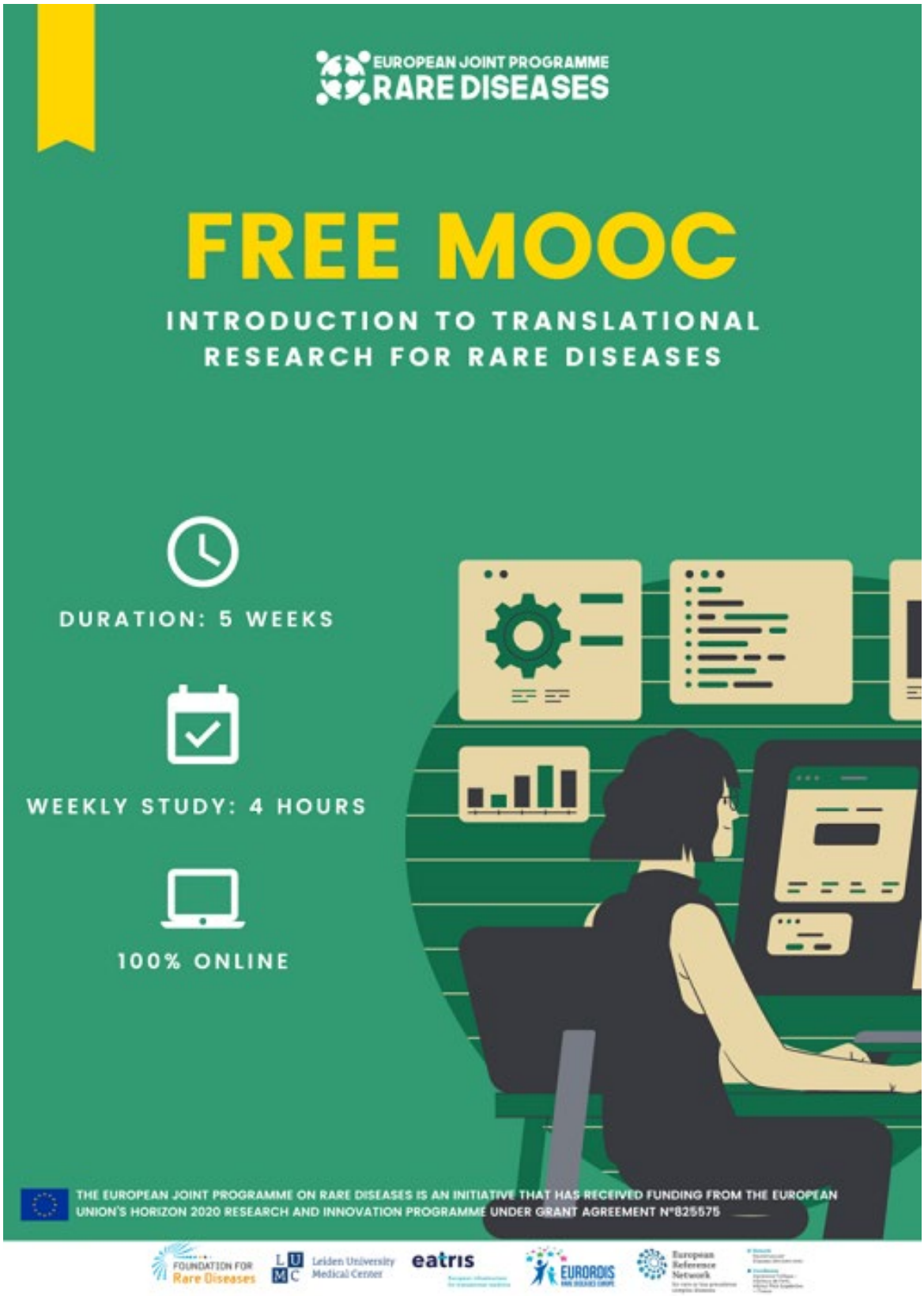
ERN GENTURIS study highlights high risk mutations associated with the development of Hereditary Diffuse Gastric Cancer related cancers

Twelve ERN GENTURIS centers participated in a study led by Carla Oliveira (Porto Comprehensive Cancer Centre) identifying the alterations in the CDH1 gene that specifically increase the risk of developing cancers associated to Hereditary Diffuse Gastric Cancer (HDGC) syndrome. This study has also defined three new clinical criteria, in addition to those currently used, which will be fundamental to identify families at risk for genetic testing, and to act prophylactically in order to prevent the development of these oncologic diseases of extremely high mortality. The study was published in the Lancet Oncology journal.
For more information, click here.



Translated lay summaries and pocket guides for the schwannomatosis and PHTS guidelines

Translations of the pocket guides and lay summaries for the ERN GENTURIS schwannomatosis and PTEN Hamartoma Tumour Syndrome (PHTS) guidelines in a number of languages are now available via https://www.genturis.eu/l=eng/Guidelines-and-pathways/Clinical-practice-guidelines.html.
The Li-Fraumeni and Heritable *TP53*-related cancer syndromes (h*TP53*rc) pocket guide and lay summary translations will follow soon.



More information about this MOOC:
https://www.futurelearn.com/courses/introduction-to-translational-research-for-rare-diseases

EJP RD - ERN workshop on psychosocial impact of rare disease

This EJP RD - ERN workshop aims to to raise awareness for the psychosocial impact of having a rare disease, and form a group of researchers in the mental well-being and social sciences domain for rare diseases.

See https://www.ejprarediseases.org/event/ejp-rd-ern-workshop-rare-together/