

Newsletter



Genetic Tumour Risk Syndromes (ERN GENTURIS)

October 2020 | www.genturis.eu

ERN GENTURIS - upcoming webinars and EJP RD fellowship exchange

The first ERN GENTURIS Webinar after the summer has been successful! Prof. Elke Holinski-Feder shared her expertise on HNPCC related tumour risk syndromes during this webinar, which was joined by over 200 attendees.

In this newsletter you will find more details on upcoming ERN GENTURIS webinars. Do not forget to register and to save the dates! Registration links for the next three webinars can be found in this newsletter. For more information about all planned webinars, please visit our website.

Also, the Research Mobility Fellowship Call of the European Joint Programme for Rare Diseases is now open. More information can be found in this newsletter. Please, share the news amongst PhD students and medical specialists in training in your ERN (affiliated) institution.



From Li-Fraumeni to Heritable TP53-Related Cancer Syndromes

14 October, 16:00 CEST - Prof. Thierry Frebourg, Rouen

Prof. Thierry Frebourg, head of the department of genetics of the Rouen University Hospital, will focus on Li-Fraumeni and heritable TP53-related cancer syndromes during this webinar.

He will discuss which patients should be tested for TP53, how germline TP53 variants should be interpreted and what the medical consequences of germline TP53 variants are. Also, the type of medical follow-up that should be offered to germline TP53 variant carriers will be discussed. If you would like to attend this webinar, please register here.

Neurocutaneous tumour syndromes

28 October, 16:00 CET - Prof. Gareth Evans, Manchester

Prof. Evans will focus on the neurofibromatoses (NF1, NF2 and schwannomatosis) and Gorlin syndrome. He will discuss the epidemiology and tumour risks and will highlight new diagnostic criteria that have recently been developed, as well as genotype phenotype correlations and mosaicism. Management including new drug treatments will also be covered. If you would like to join this webinar, please, register here.





Pheochromocytoma/ paraganglioma – benefit of combined germline and tumour testing for PPGL patients

11 November, 16:00 CET - Prof. Evelin Schröck, Dresden

Every patient afflicted with a PPGL tumour should be offered genetic testing, since germline mutations are found in 30% of all PPGL patients and in 80% of young PPGL patients. Integration of germline and tumour sequencing data greatly benefits patients by enhancing detection rates of pathogenic variants. Tumour sequencing is imperative for the identification of potential somatic driver mutations and potential exclusion of hereditary tumour syndromes. This is of importance for patients and their families. Surveillance programs and clinical management programs are needed. The establishment of in vitro models enables fast analysis of VUS in SHDB, VHL and FH (PoC). The aim is to streamline classification of VUS identified in PPGL patients, which contributes to precision cancer medicine. If you would like to join this webinar, register here.

Upcoming webinars:

14 October, 16:00 CEST:

Li-Fraumeni and heritable TP53-related cancer syndromes – Prof. Thierry Frebourg

28 October, 16:00 CET:

Neurocutaneous tumour syndromes – *Prof. Gareth Evans*

11 November, 16:00 CET:

Pheochromocytoma/paraganglioma – Prof. Evelin Schröck

25 November, 16:00 CET:

Hereditary Gastric Cancer – Prof. Carla Oliveira

9 December, 16:00 CET:

PTEN hamartoma tumour syndrome – Prof. Nicoline Hoogerbrugge

13 January 2021, 16:00 CET:

Gastrointestinal polyposis syndromes – Prof. Stefan Aretz

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

2nd Call Research Mobility Fellowship is now open! Apply before 13 November!

The call for Research Mobility Fellowships aims to financially support PhD students and medical doctors in training working in ERN-Full Member or Affiliated Partner institutes to undertake short scientific visits (secondments) fostering specialist research training outside their countries of residence and within one of the ERN-Full Member or Affiliated Partner institutes.

PhD students and medical specialists in training, who have finished their first year of training, can apply for a fellowship within this call. The duration of these short scientific visits (secondments) can be 1 to 3 months.

Applicants that are selected will receive a reimbursement for travel up to 400€ for the entire fellowship and accommodation expenses up to 2000€ per month.

For more information on the call, please

visit: https://www.ejprarediseases.org/index.php/training-and-empowerment/ern-trainings/

ERN GENTURIS Coordinating Center:

Radboud university medical center The Netherlands

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

How to unsubscribe?

You received this newsletter because you are a contact of ERN GENTURIS. If you no longer wish to receive newsletters, please unsubscribe.

© Radboudumc







