Radboudumc
university medical center

Newsletter

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

May 2021 | www.genturis.eu

Royal decoration for Nicoline Hoogerbrugge

Prof. Nicoline Hoogerbrugge, coordinator of ERN GENTURIS, is appointed by His Majesty as Knight in the Order of the Dutch Lion, a highly prestigious decoration. She is awarded the decoration because of her exceptional academic achievements in the field of hereditary cancer, which include initiating the Radboudumc Center of Expertise for Hereditary cancer and ERN GENTURIS.
See https://www.radboudumc.nl/en/news/2021/royal-decorations

Webinar 'A personal history about hereditary diffuse gastric cancer'

Tuesday 11 May 2021, 16:00-17:00 CEST

Dr. Tanya Bisseling, Radboudumc, Nijmegen

Dr. Tanya Bisseling works as a gastroenterologist in the Radboud University Medical Center Nijmegen, the Netherlands. She is one of the medical specialists of Radboudumc Center of Expertise on Hereditary Cancer.

In 2002 she was the one to hand in the clues to her colleagues to unravel the cause of gastric cancer in a few young members in her own family. As such, she herself became the first recognized carrier of a CDH1-pathogenic germline variant int the Netherlands.

In this unique webinar she will guide you over her personal path as a patient and she will complete the pavement with professional information.

If you would like to attend this webinar, please register here: https://register.gotowebinar.com/register/3464065670546269452

Webinar 'Constitutional Mismatch Repair Deficiency (CMMRD) syndrome'

Wednesday 26 May 2021, 16:00-17:00 CEST

Dr. Chrystelle Colas, Institut Curie, Paris

Doctor Chrystelle Colas is a geneticist specialized in cancer predispositions. She is the co-head of the Clinical Unit of the Genetic Department of Institut Curie in Paris, France.She has a PhD in cancer genetics. Her main research interest fields are digestive cancer predispositions, mismatch repair instability and Constitutional Mismatch Repair Deficiency. She is one of the founder members of the European Consortium “Care for CMMRD” and she is in charge of the European Database about this disease. This webinar will focus on clinical and molecular diagnosis of CMMRD as well as care and surveillance.

If you would like to attend this webinar, please register here: https://register.gotowebinar.com/register/6254750428229774605

Upcoming webinar:

9 June: Cancer risk syndromes and risk reducing surgery – Dr. Svetlana Lagercrantz, Karolinska University Hospital, Stockholm

For more information on these webinars or for registration, please visit www.genturis.eu

Free Online course 'Diagnosing Rare Diseases: from the Clinic to research and back'

Discover the role of research, clinical investigation and data sharing in diagnosing rare diseases! This online course is a joint effort from several EJP RD Work Package 16 partners, in particular ERN GENTURIS (represented by Dr Chrystelle Colas), ERN ITHACA (represented by Prof Laurence Faivre) and FFRD (Roseline Favresse).

While primarily designed for medical students and PhD/post-doc students in biomedical sciences, this course will also be of interest to Patients Advocacy Organisations' representatives, Healthcare professionals or paramedics who want to further their knowledge of rare diseases diagnosis.

More information here: https://www.futurelearn.com/courses/rare-genetic-disease

May is PHTS Awareness Month!

PTEN Hamartoma Tumour syndrome (PHTS) is a rare genetic condition that causes an increased risk for certain cancers, benign growths, and neurodevelopmental conditions.

Currently, the European INSPECT study (**In**ves**s**tigation into **P**T**E**N-related **c**ancer and pheno**t**ype) is ongoing in PHTS patients and is coordinated by Dr. Janet Vos and

Prof. Nicoline Hoogerbrugge of the Radboud university medical center in Nijmegen, the Netherlands. The INSPECT study aims to unravel the PHTS phenotype and to provide accurate cancer risks and prognoses by using information from medical records, registries and patient questionnaires.

Both patients and professionals are invited to join the INSPECT study registration for PHTS patients aged 16 years and older at https://pten.eu/en/.

ERICA kickoff meeting in May 2021

The European Rare Disease Research Coordination and Support Action consortium (ERICA) consortium will have its kickoff meeting on 27-28 May.

All 24 European Reference Networks (ERNs) are taking part in the consortium, which aims to strengthen the clinical research activities and innovation capacity of the ERNs.

See https://www.erica-rd.eu/

CPMS: IT Platform for clinical consult between ERN Members

The Clinical Patient Management System (https://cpms.ern-net.eu/login/) is a useful tool to discuss patients with a rare disease with international experts. ERN GENTURIS recurring meetings to discuss patient cases are scheduled every first and third Friday of the month from 13:30-14:30 CEST. If you are a clinician, and you have a genturis-patient that you would like to discuss with international experts: check https://www.genturis.eu/l=eng/For-clinicians/Referring-a-patient.html

ERN GENTURIS Coordinating Center:

Radboud university medical center
The Netherlands

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