Radboudumc
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# Newsletter

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

August/September 2021 | www.genturis.eu

## Webinar 'ERN GENTURIS: The EU highway for these rare diseases'

**Wednesday 15 September 2021, 16:00-17:00 CEST**

**Prof. Nicoline Hoogerbrugge, Radboudumc, Nijmegen**

Prof. Nicoline Hoogerbrugge, professor in hereditary cancer at the Radboud university medical center, Nijmegen, will be the speaker during this lecture on the European Reference Network on Genetic Tumour Risk Syndromes (ERN GENTURIS).

Genetic tumour risk syndromes are disorders in which inherited genetic mutations strongly predispose individuals to the development of tumours. ERN GENTURIS is working to improve identification of these syndromes, minimise variation in clinical outcomes, design and implement guidelines, develop registries and biobanks, support research, and empower patients. Nicoline Hoogerbrugge is the coordinator of this ERN and will show what the Network can do for patients and for

professionals.

If you would like to attend this webinar, please register here:
https://register.gotowebinar.com/register/1960594571807548430

## Webinar 'Replication Error Repair defects in cancer and cancer predisposition'

**Wednesday 29 September 2021, 16:00-17:00 CEST**

**Ass. Prof. Katharina Wimmer, Medical University Innsbruck, Austria**

Katharina Wimmer is Associate Professor at the Institute of Human Genetics at the Medical University Innsbruck, Austria. One of her research focusses is constitutional mismatch repair deficiency (CMMRD) and more broadly constitutive replication error repair (RER) deficiencies. She co-founded the European consortium "Care for CRMMRD", which aims at improving diagnosis and management of children/young adults with this syndrome.

This webinar will (i) render an overview on the cellular effects and the therapeutic implications of RER deficiency in tumours and (ii) our current knowledge on phenotypic differences and overlap between constitutional polymerase proofreading deficiency and CMMRD. (iii) Differences between somatic and germline replication DNA polymerases POLE (POLε) and POLD1 (POLδ) pathogenic variants as well as strategies and challenges of POLE and POLD1 variant classification will be discussed.

If you would like to attend this webinar, please register here:
https://attendee.gotowebinar.com/register/3419337542797150224

## Hereditary cancers: diagnosed patients are only the tip of the iceberg

Although knowledge on pathogenic germline variants has improved over the years, researchers are hunting for new genetic causes of cancer to better identify families at higher risk to develop the disease. Read the interview with ERN GENTURIS coordinator Nicoline Hoogerbrugge in the latest edition of ESMO Perspectives:
https://perspectives.esmo.org/latest-edition/featured-content/hereditary-cancers-diagnosed-patients-are-only-the-tip-of-the-iceberg

## Solve-RD publications on new approach to solve unsolved rare diseases

For the first time in Europe hundreds of rare disease experts team up to actively share and jointly analyze existing data from unsolved rare disease patients. The Solve-RD project published a series of papers in the European Journal of Human Genetics describing the new approach and the structures established to warrant best exchange of expertise. Involved in the project are data scientists and genomics experts as well as expert clinicians and geneticists from ERNs, among which ERN GENTURIS.
For more information and a link to the papers, see https://www.medizin.uni-tuebingen.de/en-de/das-klinikum/pressemeldungen/378?press_str=

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## Newsletter ERN Exchange Programme

The ERN Exchange Programme is designed to share knowledge and stimulate collaboration between healthcare professionals in the European Reference Networks (ERNs). In this newsletter, an update is given on where the Exchange programme stands and information about the EU Digital COVID Certificate is provided that will greatly facilitate implementation of the programme. Also an impression is given of the first exchange visits that are taking place.
https://www.genturis.eu/l=eng/Assets/20210706_ERN_Newsletter-1972.pdf

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## Hereditary cancer portal launched in Portugal

The Working Group dedicated to Hereditary Cancer of the Portuguese Oncology Society (SPO) has presented the National Programme of Genetics and Cancer (PROGO) as well as the launch of an informative and educational portal on hereditary cancer. Dr Carla Oliveira, national coordinator of ERN GENTURIS: “Among others, PROGO aims to create a national structure necessary for a network organisation, allowing a better integration of Portugal in ERN GENTURIS. The portal will serve as a platform for the dissemination of the most recent guidelines, including those developed and accepted at European level".

For more information, see www.cancrohereditario.pt

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## Upcoming webinars:

Wednesday 13 October: Iosune Baraibar (Vall d'Hebron UH, Barcelona) - Update in therapeutic options for Lynch-associated colorectal cancer patients

Thursday 21 October: Jolanda Schieving (Radboudumc, Nijmegen) – PTEN hamartoma tumour syndrome in childhood

Wednesday 10 November: Rianne Oostenbrink (Erasmus MC, Rotterdam) – NF1 in children

Wednesday 24 November: Stefan Aretz (University Hospital Bonn) – Somatic Mosaicism in Tumour Genetics

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

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