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

Newsletter February 2024

ERN GENTURIS news

Rare disease day 29 February 2024

Today, on **Rare Disease Day 29 February 2024**, we come together to raise awareness and celebrate the strength and resilience of those affected by rare diseases. At the heart of our efforts lies a commitment to drive positive change, empower patients, and advance the quality of care for the rare disease patients.
ERN GENTURIS is working to improve identification of the genetic tumour risk syndromes, to minimise variation in clinical outcomes, to design and implement EU-guidelines, develop the GENTURIS registry, support research, and empower patients. The network is educating the public and healthcare professionals via the website www.genturis.eu, by organising regular webinars and courses, and by fostering sharing of best practice across Europe. Access to multidisciplinary care (digital as well as face-to-face) will be improved, to share and discuss complex cases. The network is enhancing the quality and interpretation of genetic testing.
Together, we can turn awareness into action and create a world where every person affected by a rare disease receives the care, support, and hope they deserve.

Check out the ERN GENTURIS video: here

Read the **Statement by the EU Health Commissioner Kyriakides on the World Rare Disease Day - Stronger together**

The ERN GENTURIS Task Force leaders

Task Force 4 new chair

The new chair of **Task Force 4 - Continuous education, training and development, Assist. Prof. Mateja Krajc, MD, PhD,** is a specialist in clinical genetics and public health She graduated the Medical school, University of Ljubljana, Slovenia in 1998. She obtained a PhD degree in hereditary breast cancer in January 2014 from the Vrije Universiteit Brussel, Belgium. Dr. Krajc is currently employed at the Institute of Oncology in Ljubljana Slovenia and is heading the Cancer Genetics Clinic at the Institute and runs the multidisciplinary team for hereditary cancer. She is involved in cancer genetic screening, cancer genetics research and stratified cancer screening organisation and research.
Assist.prof Krajc is also the national coordinator for Slovenia in ERN GENTURIS.

Task Force 5 new chair

The new chair of **Task Force 5 - Research, data registries, biobanking and funding, Assist. Prof. Karin Wadt, MD, PhD,** is a specialist in clinical genetics, graduated from University of Copenhagen in Denmark 1999. She obtained a PhD degree in hereditary melanoma at University of Copenhagen. Dr. Wadt is currently employed at the Clinical Genetics Department at the University Hospital Rigshospitalet in Copenhagen and is heading a Cancer Genetics Clinic with a multidisciplinary team for hereditary cancer. Dr Wadt is involved in cancer genetic screening, clinical guidelines, organisation of cancer genetics and clinical surveillance. Dr. Wadt is involved in cancer genetics research with a specific focus on childhood cancer and rare cancer syndromes.
Assist.prof Karin Wadt is also the national coordinator for Denmark in ERN GENTURIS.

ERN GENTURIS knowledge

Clinical Practice Guideline

The newly endorsed clinical practice guideline for **the diagnosis and surveillance of BAP1 tumour predisposition syndrome is available** on our website: **here.**

Publications

The recent publication in collaborative effort with the ERN GENTURIS partners was published on 13 February 2024: **The heterogeneous cancer phenotype of individuals with biallelic germline pathogenic variants in CHEK2**
Check our website: **here**

Education opportunities

ERN GENTURIS educational webinars

13 March 2024
Hereditary tumour syndromes in gastroenterology
Verena Steinke-Lange

27 March 2024
TP53 variant classification
Thomas van Overeem Hansen

More on our website:
Webinars (genturis.eu)

FREE online course - Diagnosing Rare Diseases: from the Clinic to Research and Back

In the context of the European Joint Programme for Rare Diseases EJPRD, **co-developed by Chrystelle Colas (ERN GENTURIS), Laurence Faivre (ERN ITHACA), and the Foundation For Rare Diseases,** the **Massive Open Online Course (MOOC)** addresses the diagnostic research progress, types of genetic tests for rare diseases, and the impact of having or lacking a diagnosis on patients' lives.You will be able to discover the role of **research, clinical investigation and data sharing** in diagnosing rare diseases.

The MOOC is continuously open for enrollment and the access to the content is free for the first 5 weeks.

For more information check the link

General news

Horizon Europe 'Health' single-stage calls for proposals 2024

HaDEA has published a set of new calls under Cluster 1 'Health' of Horizon Europe.

The deadline to apply to these calls is on **11 April 2024, 17:00 CEST**

EU4Health call for tenders for a study on the quality of life of **cancer patients and survivors in Europe**.

Find out more **here**
The deadline to apply is on **1 March 2024, 16:00 CET**

Rare diseases: still on the fringes of universal health coverage in Europe

A recent publication in The Lancet Regional Health – Europe February 2024 by **Birutė Tumienė (ERN GENTURIS), Augutė Juozapavičiūtė and Vytenis Andriukaitis,** addresses strategies for achieving universal health coverage for rare diseases in Europe. The article advocates for Centres of Excellence,

efficient care pathways, research collaboration, and patient-centred care, aiming to provide equitable healthcare for Europeans with rare diseases.

Funding opportunities

European Commission Calls

- HaDEA Calls for **Proposals on Health**
- HaDEA Calls for **Tenders on Health**
- Horizon Europe calls for **Funding on Health**

Upcoming Meetings & Events

National Integration of the ERNs: Kick Off Meeting

JARDIN (the Joint Action on the integration of European Reference Networks (ERNs) into national health systems) kick-off meeting will be held on on 6-8 March 2024.

On 8 March (8:30 - 17:30 CET), online attendees can join a session on Rare Diseases in the EU: Joint Action shaping the future of ERNs held in cooperation with the **European Economic and Social Committee, Commissioner for Health and Food Safety, Stella Kyriakides** will give a keynote speech.

To register go here

12th European Conference on Rare Diseases (ECRD) will be held in Brussels and online on 15-16 May 2024. The deadline for submitting poster abstracts for ECRD 2024 has been extended to **4th March 2024!**
Researchers, patient groups, healthcare professionals and any other interested parties in the rare disease community are invited to submit their **poster abstract** (maximum of 300 words, in English only).

Learn more and submit your abstract now: Posters - ECRD2024 (rare-diseases.eu)
Find more info here

EJP RD Final Conference 27-28 May 2024

The EJP RD Final Conference, will be held in Bari, Italy from 27-28 May 2024 for invited people and will be open for all online .

The conference is set to bring together leading experts, professionals, and enthusiasts from around the world. Find more about the event **here.**

Registration deadline: 31 March 2024 - **here**

Pre-ESHG ERN GENTURIS meeting in Berlin, Germany

The next ERN GENTURIS meeting will take place prior to the ESHG conference in Berlin on 31 May and 1 June 2024. ERN GENTURIS members who attend the ESHG conference are invited to participate.

SAVE THE DATE: 31 May -1 June 2024

5th course in Hereditary Cancer Genetics, Bertinoro, Italy

This course, organised by members of the ERN GENTURIS, aims to deliver up-to-date knowledge on hereditary cancers to clinical and molecular geneticists in training or certified. The programme is available here and the registration will open soon. More information will follow.

SAVE THE DATE: 17-20 September 2024

ERN GENTURIS

With every diagnosis we can help an entire family

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