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Newsletter

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

December 2021 | www.genturis.eu

Sirkku Peltonen Pierre Wolkenstein

Webinar 'Cutaneous neurofibromas'

Thursday 27 January 2022
16:00-17:00 CET

Dr Sirkku Peltonen, University of Gothenburg, Sweden and Prof. dr. Pierre Wolkenstein, University Hospital Henri-Mondor Hospital, Paris, France

Sirkku Peltonen, MD, PhD, is a dermatologist and has led the NF1 clinic in Turku University Hospital, Turku, Finland since 1999. Her studies on NF1 mainly consider the biology of cutaneous neurofibromas, but she has also published on translational and clinical topics related to NF1 such as speech, osteoporosis and craniofacial features. Her most recent publications are epidemiological studies on NF1 concerning incidence, prevalence, mortality, cancer, malformations, other co-morbidities, and pregnancies. At present she works as a Professor of Dermatology and Venereology in University of Gothenburg, Sweden.

Pierre Wolkenstein is Professor at the Faculty of Medicine and Health of Paris Est. He is dermatologist and oncologist, and head of the Department of Dermatology at Henri-Mondor Hospital. Since the 1990s, he has been coordinating the management and clinical research of neurofibromatoses. His team performed the first face transplant on a patient with neurofibromatosis 1, identified the first modifying gene, conducted therapeutic trials, and developed basic research on NF1 animal models. He is president of the European Neurofibromatoses Group.

Cutaneous neurofibromas are the hallmark of NF1 and their number in the visible body areas is related to quality of life in adults with NF1. In this webinar cutaneous neurofibromas will be discussed from several points of view: their pathogenesis, clinical picture, impact on patients' lives and treatment options.

If you would like to attend this webinar, please register here: https://register.gotowebinar.com/register/5507349709609624589

Registration for the ERN GENTURIS 5-year anniversary conference on 10-11 Feb 2022 is still open!

On 10-11 February 2022, ERN GENTURIS will celebrate its 5-year anniversary with a free online conference.

The conference will feature updates on the ERN GENTURIS tumour predisposition syndromes, reports on special ERN GENTURIS projects and patient perspectives. It will also include a half-day introductory educational session specially aimed at general practitioners.

For the conference program and the registration links, visit https://www.genturis.eu/l=eng/News-and-events/ERN-GENTURIS-conference-Feb-2022.html

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ERN GENTURIS special issue of the European Journal of Medical Genetics

Another 3 papers were recently published in the EJMG ERN GENTURIS special issue, bringing the total to 7 papers, with more papers to follow! See https://www.sciencedirect.com/journal/european-journal-of-medical-genetics/special-issue/10PFC6MB478

ERN GENTURIS welcomes 25 new Full Members

The Board of Member States has approved 620 applicants for full membership of the existing ERNs, including 25 new Full Members for ERN GENTURIS. The new memberships officially started on 1 January 2022. You can find the list of the new members sorted by ERN here: https://ec.europa.eu/health/sites/default/files/ern/docs/call2019_decision_ern_en.pdf

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First GENTURIS registry meeting for participating centres

On 12 January, a successful GENTURIS registry meeting was held for the ERN GENTURIS network with 77 representatives attending, including from new Full Member centres. Janet Vos and Bianca Hilhorst from Radboudumc Nijmegen elaborated on the legal and ethical aspects and the data collection procedure of the registry. Rolf Sijmons from UMC Groningen explained the data access procedure.
For more information about the GENTURIS registry, see https://genturis-registry.eu/

The European Conference on Rare Diseases 2022

EURORDIS and Orphanet organise the European Conference on Rare Diseases 2022. ECRD 2022 will take place fully online and is spread over 5 half days, from 27 June to 1 July 2022. Registration will open on 7 March 2022.

Poster abstracts submissions for the ECRD 2022 are now open until 31st March 2022. EURORDIS-Rare Diseases Europe is offering patient fee waivers for up to 80 patients' advocates to attend the ECRD. Applications deadline: 31 January 2022.
See for more information: https://www.rare-diseases.eu/.

ERN-EYE joint webinar with ERN GENTURIS on Neurofibromatosis 1 & 2

ERN-EYE organises a joint webinar with ERN GENTURIS on Neurofibromatosis 1 & 2, on Tuesday 3 February 2022, 18.30 pm - 19.30 pm CET.

- ERN GENTURIS members Eric Legius and Kathleen Claes will discuss the new diagnostic criteria, molecular aspects of NF-1 and NF-2 and genetic testing.
- ERN GENTURIS ePAG Claas Röhl will focus on patient testimonies.
- ERN-EYE members Dominique Brémond-Gignac and Matthieu Robert will present Ophthalmological aspects, retinal imaging & OCT.

For more information and registration, see
https://www.ern-eye.eu/ern-eye-webinars/3-february

Upcoming webinar:

Wednesday 23 February 2022:
Marc Tischkowitz (University of Cambridge, UK)
Polygenic Risk Scores in breast and ovarian cancer risk prediction – ready for the clinic?

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

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