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Newsletter

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

December 2021 | www.genturis.eu

Webinar 'Legius syndrome and its link with Neurofibromatosis type 1'

Tuesday 7 December 2021, 16:00-17:00 CET

Prof. dr. Eric Legius, KU Leuven, Belgium

Eric Legius is professor in Human Genetics at the University of Leuven in Belgium and current head of the Department of Clinical Genetics at the University Hospital of Leuven. He is a clinician scientist. His research is targeted towards Neurofibromatosis type 1 and related conditions. The research group contributed successfully towards the understanding of the molecular aetiology of a number of tumours in NF1. In 2007 his research team identified a new condition resembling Neurofibromatosis type 1, now known as Legius syndrome.

Legius syndrome is an autosomal dominant condition characterized by multiple café-au-lait spots and caused by heterozygous pathogenic variants in SPRED1. The SPRED1 protein is an important regulator of the ras-mapk pathway and the protein is needed to recruit neurofibromin (the protein produced by the NF1 gene) to the cell membrane where neurofibromin can downregulate activated ras. Legius syndrome and Neurofibromatosis type 1 are both characterized by multiple café-

au-lait spots and the two conditions are very similar in children.

Recently diagnostic criteria for Legius syndrome and revised diagnostic criteria for Neurofibromatosis type 1 were published stressing the importance of molecular testing to differentiate the two diseases that affect the Ras-MAPK pathway in a similar way.

We will discuss the binding of SPRED1 to neurofibromin and the effect of missense variants in these regions in both genes on the function of the respective proteins. An important difference between the two diseases is the absence of Neurofibromatosis type 1 related tumours in Legius syndrome. A similarity is the effect on cognition and behaviour.

If you would like to attend this webinar, please register here:
https://register.gotowebinar.com/register/7934968027657104142

Registration is open - ERN GENTURIS 5-year anniversary conference on 10-11 Feb 2022

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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Upcoming webinars:

Thursday 27 January 2022:
Sirkku Peltonen (Univ. Gothenburg, Sweden) & Pierre Wolkenstein (Henri-Mondor Hospital, France)
Cutaneous neurofibromas

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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