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

Newsletter September 2023

ERN GENTURIS news

ERN GENTURIS

With every diagnosis
we can help an entire family

WOW!

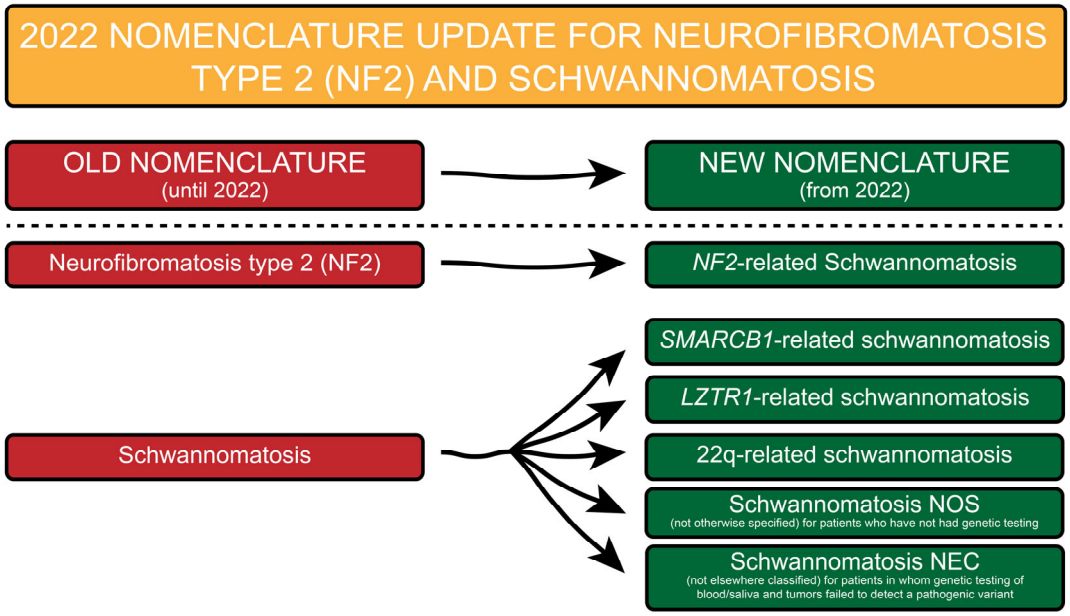
Evaluation Results

ERN GENTURIS as a Network received in its final report a **100% score.** All ERN GENTURIS Full Members that were approved in **2017** went through the **Independent Evaluation Body** evaluation as specified in the Commission Delegated Decision 2014/286/EU and

Implementing Decision 2014/287/EU. Final Reports have been shared with the Network and with our Members, and will be shared with the ERN Board of Member States.

Read the full news **here**
The evaluation process is explained on the Commission website: **here**

New nomenclature



Thematic Group 1 Neurofibromatosis recently published a **statement** about adoption of new nomenclature. The TG1 leader**, Ignacio Blanco,** explains:

"Now it is the time to embrace the new nomenclature (Neurofibromatosis type 2 will become NF2-related schwannomatosis, and schwannomatosis will be categorised as SMARCB1-related schwannomatosis, LZTR1-related schwannomatosis, 22q-related schwannomatosis, or schwannomatosis not otherwise specified (NOS)) within ERN GENTURIS. We encourage all GENTURIS members to advocate for the adoption of this nomenclature in their respective countries."

Letter in the Guardian: The UK is missing out on EU collaboration to improve treatment for rare diseases

A letter from ERN GENTURIS Supporting Partner **Prof Marc Tischkowitz** from Cambridge University Hospital on the importance of **UK re-joining the ERNs** was published in the Guardian on 21 September 2023.

"The absence of an agreement on UK participation in ERNs has negated years of progress made by UK clinicians, researchers and patient advocates, and it has diminished our ability to provide the best care for the millions of children and adults with rare diseases and

complex conditions. We now urgently need to make up for the time we have lost."

Read the whole letter here: [The UK is missing out on EU collaboration to improve treatment for rare diseases | Brexit | The Guardian]

The ESMO Preceptorship on Hereditary Cancer Genetics, 29-30 September 2023 in Paris, France

The course is co-chaired by **ERN GENTURIS members Judith Balmaña and Nicoline Hoogerbrugge**. In this **accredited educational course** 43 specialists from 21 countries participate (33 of them are medical oncologists). The majority of the lecturers are **ERN GENTURIS** members: Stefan Aretz (Germany), Matteo Lambertini (Italy), Marjolijn Ligtenberg (the Netherlands), Carla Oliveira (Portugal), Rolf Sijmons (the Netherlands), Marc Tischkowitz (United Kingdom). The main objectives of the course are:

- to learn about the best clinical practice in the multidisciplinary management of hereditary cancer syndromes
- to understand the molecular basis and heterogeneity of the genetic susceptibility to develop cancer
- to learn how to recognise, diagnose, treat and provide prevention recommendations to patients with a germline genetic susceptibility

Education opportunities

Diagnosing Rare Diseases: from the Clinic to Research and Back

Would you like to discover the role of **research, clinical investigation and data sharing** in diagnosing rare diseases? Join the 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) and Laurence Faivre (ERN ITHACA), the Foundation For Rare Diseases,** this **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.

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

Upcoming ERN GENTURIS educational webinars

11-10-2023
Haematological malignancies – a genetic counselling perspective
Bev Speight

25-10-2023
Peutz-Jeghers syndrome – the clinical genetic perspective
Anja Wagner & Esther Korpershoek

For more information visit www.genturis.eu

General ERN news

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

Conference on Rare Diseases and the ERNs, 10-11 October

The Spanish Presidency of the EU Council is organising a **conference on rare diseases and the ERNs** from **10-11 October 2023** in Bilbao, Spain. The conference will focus on strategies for overcoming the main challenges facing the EU policy framework on rare diseases and ERNs.

Conference on the NF Best Practice, 15 October

NF Patients United is hosting the **NF Best Practice 2023** on October 15, 2023 at 09h30 to 15h15 CEST. The meeting will be held in Rotterdam, the Netherlands. **ERN GENTURIS** NF expert, **Rianne Oostenbrink** will give research updates about EU PEARL: EU-Patient Centric Clinical Trial Platforms. Two Patient Representatives will update about two guidelines:

- **ERN GENTURIS** tumour surveillance guidelines for individuals with neurofibromatosis type 1 (Claas Roehl)
- **ERN GENTURIS** clinical practice guidelines for the diagnosis, treatment, management and surveillance of people with schwannomatosis (Melpo Pittara)

All talks are aimed at patients with NF1, NF2-related Schwannomatosis and Schwannomatosis, their caregivers as well as for all interested.
Here is more information: **link**

European Cancer Summit, 15-16 November

The **European Cancer Summit 2023** will be held in Brussels, Belgium and online on **15-16 November 2023** with the theme "**Accelerating Momentum: A Manifesto to 2030**". Commissioner Stella Kyriakides has confirmed her in-person participation.

ERICA & EJP RD Joint Conference, 21 November

The **ERICA & EJP RD Joint Conference** will be held in Amsterdam, the Netherlands on **21 November 2023** (with a pre-conference dinner on 20 November 2023).
The meeting will provide a closer overview of all the relevant activities and joint efforts in the field of **rare disease research** during the lifetime of **EJP RD** and **ERICA** with a further focus on the future **Rare Diseases Partnership** and **ERN research activities**.

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