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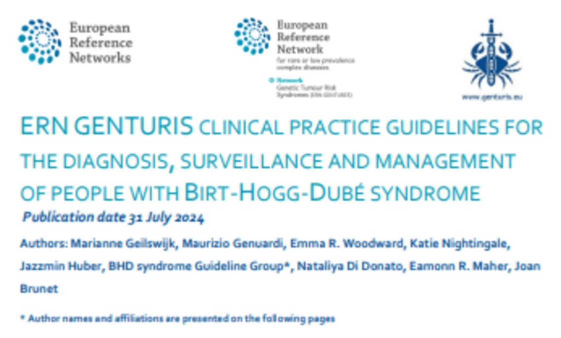
ERN GENTURIS Newsletter

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

July & August 2024

ERN GENTURIS news

ERN GENTURIS clinical practice guidelines: Birt-Hogg-Dubé syndrome



European Reference Networks
European Reference Network
www.genturis.eu

ERN GENTURIS CLINICAL PRACTICE GUIDELINES FOR THE DIAGNOSIS, SURVEILLANCE AND MANAGEMENT OF PEOPLE WITH BIRT-HOGG-DUBÉ SYNDROME

Publication date 31 July 2024

Authors: Marianne Geilswijk, Maurizio Genuardi, Emma R. Woodward, Katie Nightingale, Jazzmin Huber, BHD syndrome Guideline Group*, Nataliya Di Donato, Eamonn R. Maher, Joan Brunet

* Author names and affiliations are presented on the following pages

EUROPEAN REFERENCE NETWORKS
FOR RARE, LOW PREVALENCE AND COMPLEX DISEASES
Share. Care. Cure.

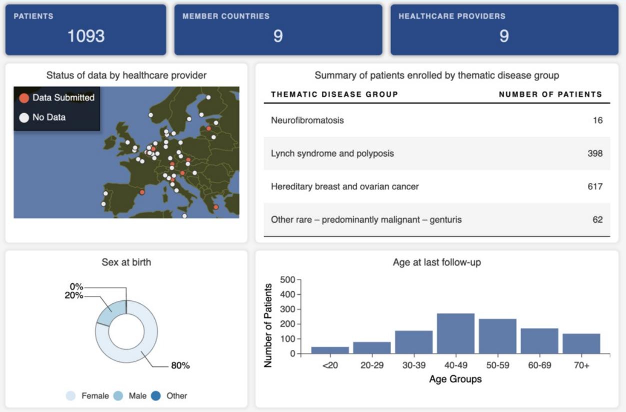
The ERN GENTURIS clinical practice guidelines for the diagnosis, surveillance and management of people with **Birt-Hogg-Dubé syndrome** were published on 31 July 2024.

We would like to thank all the authors, as well as **ERN-LUNG, ERKNet** and **EURACAN** for reaching out to their members to participate in the Delphi, and all Delphi participants for their participation in the Delphi survey.

To download the **complete guideline, the EJHG publication, the pocket guide and the plain language summary in English,** please go to the ERN GENTURIS website:

website

ERN GENTURIS Registry



PATIENTS 1093

MEMBER COUNTRIES 9

HEALTHCARE PROVIDERS 9

Summary of patients enrolled by thematic disease group

THEMATIC DISEASE GROUP	NUMBER OF PATIENTS
Neurofibromatosis	16
Lynch syndrome and polyposis	398
Hereditary breast and ovarian cancer	617
Other rare – predominantly malignant – genturis	62

Status of data by healthcare provider
Data Submitted
No Data
Sex at birth
0%
20%
80%
Female
Male
Other
Age at last follow-up
Number of Patients
500
400
300
200
100
0
<20
20-29
30-39
40-49
50-59
60-69
70+
Age Groups

The GENTURIS registry is a web-based platform that facilitates standardized data registration and sharing of data across Europe to improve diagnostics, treatment and disease prevention in patients with a genetic tumour risk syndrome (genturis).

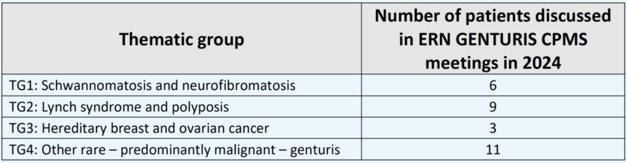
The registry enables a new wave of **research opportunities** from translational to epidemiological studies to clinical trials, and will accelerate the generation of new knowledge on genetic tumour risk syndromes.

- Since the last newsletter (June 2024) more than **276** new patients have been included in the GENTURIS registry.
- The majority of the patients enrolled last month originate from **Spain.**

For more information, click the link below or contact the UMCG team via **genturis-registry@umcg.nl**

more information

ERN GENTURIS overview of patients discussed in the CPMS in 2024



Thematic group	Number of patients discussed in ERN GENTURIS CPMS meetings in 2024
TG1: Schwannomatosis and neurofibromatosis	6
TG2: Lynch syndrome and polyposis	9
TG3: Hereditary breast and ovarian cancer	3
TG4: Other rare – predominantly malignant – genturis	11

In the discussions organized through the Clinical Patient Management System (CPMS), ERN GENTURIS has provided tailored expert advice to **29** genturis patients and their families in 2024 so far. For more information on how to refer a patient for discussion in an ERN GENTURIS CPMS meeting and how to use the CPMS, please see:

- How to refer a patient
- CPMS information and policy
- CPMS guides

CPMS 2.0 information

Please visit our website for a detailed overview of all information regarding the newly developed CPMS (CPMS 2.0).

If you are willing to help test CPMS 2.0, please follow the instructions included on the CPMS 2.0 information webpage linked above.

Publications

The recently published article: "**Model matchmaking via the Solve-RD Rare Disease Models & Mechanisms Network (RDMM-Europe)**" explains the importance of establishing the RDMM-Europe and its international integration into the worldwide RDMM network for achieving the aims of Solve-RD, which are to find novel disease genes and define disease mechanisms for ultra-rare diseases.

full article

Education opportunities

ERN GENTURIS educational webinars

25 September 2024
ERN GENTURIS BHD syndrome guidelines
Marianne Geilswijk

23 October 2024
Adjuvant therapy options for hereditary breast cancer

More on our website:

webinars

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

course

Pre-InSiGHT meeting for patients and patient representatives with Lynch syndrome or polyposis

The Pre-InSiGHT meeting for patients and patient representatives with Lynch syndrome or polyposis took place on June 18th 2024. This meeting was specifically dedicated to patients with hereditary colorectal cancer, Lynch syndrome or polyposis, and was organised by the International Society for Gastrointestinal Tumours (InSiGHT) in close collaboration with ERN GENTURIS. It was held online on the day before the start of the InSiGHT scientific conference in Barcelona.

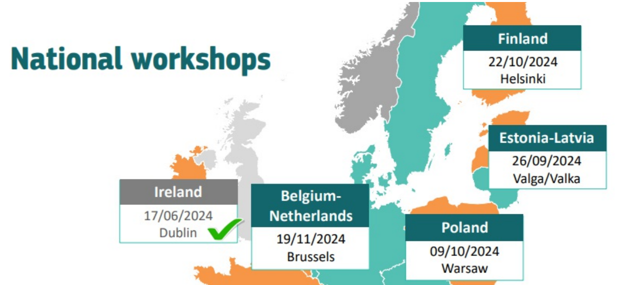
Watch the recordings:

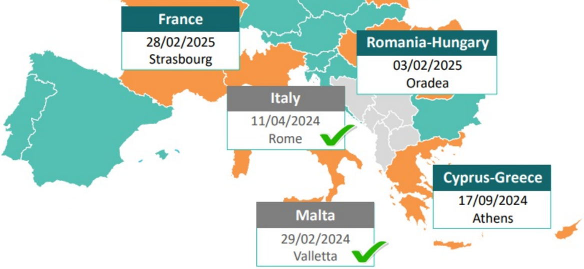
recordings

GENERAL NEWS

JARDIN a Joint Action (JA) created to achieve the integration of European Reference Networks (ERNs) into National Healthcare Systems has recently launched the website.

This initial version of the website has been developed to provide comprehensive information about the Joint Action, including its objectives, mission and vision, work packages, and ERNs.


National workshops
Finland
22/10/2024
Helsinki
Estonia-Latvia
26/09/2024
Valga/Valka
Ireland
17/06/2024
Dublin
✓
Belgium-Netherlands
19/11/2024
Brussels
Poland
09/10/2024
Warsaw


France
28/02/2025
Strasbourg
Romania-Hungary
03/02/2025
Oradea
Italy
11/04/2024
Rome
✓
Cyprus-Greece
17/09/2024
Athens
Malta
29/02/2024
Valletta
✓

The Joint workshop between **Greece and Cyprus** on patients' rights in cross-border healthcare and European Reference Networks will be held in Athens, on **17 September 2024**. The workshop will be in Greek, with English interpretation, and you have the option to attend it online.

Please register through the following link

The next workshop (joint workshop between Estonia and Latvia) will be held in **Valga, Estonia, on 26 September**. Invitation, draft agenda, and more information will follow soon.

Funding opportunities

European Commission Calls

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

EU4Health call for tenders

CALL FOR TENDERS
Study on the provision of care for adolescent and young adult cancer patients
#HaDEA #EU4Health #HealthUnion
European Commission
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HaDEA has published the EU4Health call for tenders HADEA/2024/OP/0027 - Exploratory study on the provision of care for Adolescent and Young Adult (AYA) cancer patients in the Union.
The aim of this study is to map the provision of healthcare to Adolescent and Young Adults (AYA) cancer patients and survivors in all EU countries, Norway and Iceland.

Find out more:

here

Upcoming Meetings & Events

5th course in Hereditary Cancer Genetics

This course, organized by members of **ERN GENTURIS**, aims at delivering up-to-date knowledge on hereditary cancers to clinical and molecular geneticists in training or certified. It creates the best opportunity for interaction and discussion with experts from all over Europe, in the fabulous environment of Bertinoro, the headquarters of the ESHG sponsored courses. The faculty combines experts from many fields of cancer genetics known for their didactic skills. Participants are encouraged to present a clinical or genetic case in a Poster format for on-site discussion.
The course will take place on 17-20 September 2024.

more information

Hereditary cancer and genetic counselling

HELSE BERGEN
Haukeland University Hospital

HEREDITARY CANCER AND GENETIC COUNSELLING IN THE NORDIC COUNTRIES

Skien, Norway

November 12, 2024

First meeting about **Hereditary cancer and genetic counselling in the Nordic countries** will take place on **November 12th** in Skien, Norway. The meeting is open to all healthcare professionals working with hereditary cancer.

Deadline for registration is October 1st, 2024 .

Abstracts:
You can choose between two categories:

1. Scientific project: submit a structured abstract describing your study. The abstract should include names of all authors and their affiliations. The name of the person presenting the abstract must be underlined. Font: Times New Roman, size: 12; Maximum length: 250 words (excluding title, authors, and affiliations).

2. Challenging clinical case: submit a PowerPoint presentation on a challenging case you want to discuss with our Nordic expert panel. The presentation should be of maximum 2-3 slides (1 slide pedigree, 1-2 slides pertinent details). In the e-mail you should include a title of your presentation, your name, place of work, and position. Junior colleagues will be prioritized for the case presentations.

Send your abstract/PPT to: cathrine.bjorvatn@helse-bergen.no
Deadline for abstract submission is October 1st .

registration

For an EU commitment to tackle rare diseases

For an EU commitment to tackle rare diseases
Az EU elkötelezettsége a ritka betegségek elleni küzdelem mellett
29.11.2024 | Budapest | #RareDiseases
European Economic and Social Committee
HU24EU
HUNGARIAN PRESIDENCY OF THE COUNCIL OF THE EU 2024

The European Economic and Social Committee (EESC), under the auspices of the Hungarian Presidency of the EU Council, is organising a major conference entitled "For an EU commitment to tackle rare diseases" on 29 November 2024 from 9:00 am to 4:30 pm.

more information

ERICA GA & ERN Research Conference

Save the Date:
ERICA General Assembly &
ERN Research Conference
11-13 December 2024 in Udine, Italy
ERICA
European Reference Network
MetabERN

The European Rare Disease Research Coordination and Support Action consortium (ERICA) 4th General Assembly & ERN Research Conference will take place **11-13 December, 2024** in Udine, Italy.

This event is hosted by MetabERN.

There will also be a Call on Abstracts related to ERN Research.

more information

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Website and LinkedIn

ERN GENTURIS
With every diagnosis we can help an entire family

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