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

GENTURIS registry

Newsletter special edition - GENTURIS registry

June 2023| http://genturis-registry.eu/

Preface

This special edition newsletter is in celebration of the establishment of the GENTURIS registry. Over the last years the GENTURIS registry has been established and the implementation of the registry is now up and running. This newsletter shows an impression of the GENTURIS registry and gives a tour along all developments.

Introduction of the GENTURIS registry

The Genetic Tumour Risk Syndrome (GENTURIS) registry is affiliated to ERN GENTURIS. The aim of the GENTURIS registry is to enable research that will provide insights into the natural disease history and care pathway by gathering data from a large number of genturis patients and providing regulated data access following a data access policy.
The GENTURIS registry is built on the Molgenis data platform which aligns with the FAIR principles. The registry is designed as a central database. Each of the centres have their own data space in which the patient-level data is only accessible to the respective centre.

Governance of the GENTURIS registry

During the establishment phase of the GENTURIS registry, a protocol, minimal datasets, patient information, handbook, data access policy, and collaboration agreement were developed within the framework of ERN GENTURIS.
All centres participating in the GENTURIS registry fulfil all legal and ethical requirements before accessing and enrolling patients into the live GENTURIS registry. For training purposes, a test environment of the registry is available. Access can be requested through the centre's representative(s).

For more information, please visit the GENTURIS registry website.

Enrolment in the GENTURIS registry

The GENTURIS registry includes genturis patients with a proven pathogenic variant and highly suspected patients without a genetic diagnosis. The registry covers the disease groups of ERN GENTURIS: 1) Neurofibromatosis; 2) Lynch Syndrome and polyposis; 3) Hereditary breast and ovarian cancer; 4) other rare and predominantly malignant genturis.
The first patients were enrolled in the beginning of 2023. The local implementation of the GENTURIS registry is up and running, and more and more centres are actively enrolling patients. The dashboard on the GENTURIS registry website provides a live overview of summary statistics including disease group, age, gender and country distribution.

Visit the Dashboard | GENTURIS Registry (genturis-registry.eu) for today's statistics.

Cross ERN collaboration

Across Europe all 24 European Reference Networks (ERNs) are setting up patient registries. As the registries have many similarities, cross-ERN collaboration and alignment is important. This is supported through the European joint program of rare diseases (EJP-RD) and the European Rare Disease Research Coordination and Support Action consortium (ERICA). As part of this, for instance, a common minimal dataset and a template for ERN registry patient information were developed, and a team of FAIR data stewards connect with each of the ERN registry teams.

For more information, please visit EJP RD – European Joint Programme on Rare Diseases (ejprarediseases.org) and ERICA | The European Rare Disease Research Coordination and Support Action (erica-rd.eu)

Interoperability of the GENTURIS registry

When we want to learn more about rare genetic tumour risk syndromes, connecting data from multiple sources increases our possibilities to understand the disease. The **FAIR principles** (Findable, Accessible, Interoperable, Reusable) help us do this by making data easy to find, access, and use. This includes encouraging data discovery, global identifiers, common nomenclature and rich metadata. Interoperable data means that machines can connect and query different datasets without additional help from people, and governance models can be set in place to regulate the access, for instance to metadata or high-level aggregate data and to specific user profiles.
The GENTURIS registry is built on the MOLGENIS data platform, which uses special templates to store and retrieve data in an interoperable way. ERN GENTURIS is embracing FAIR to further enhance data quality and maximize the future potential of data for research and clinic. In this context, the GENTURIS registry minimal and disease-specific dataset is based on recognised ontologies and nomenclatures (e.g. ICD-10, ICD-O, HPO, OMIM, HUGO). In addition, the registry is listed in resource

finders such as Orphanet and the European Platform on Rare Disease Registration Directory of Registries (ERDRI.dor) and the metadata repository (ERDRI.mdr).

GENTURIS registry FAIR Data Point

A relatively simple way to increase FAIRness is a FAIR Data Point. This is a document that machines can read, containing linked data that describes an online resource and its available datasets. By creating a FAIR Data Point, the resource becomes easier to find and access. The resource becomes findable by informing another machine or central search index of its existence, after which the contents of the FAIR Data Point are automatically harvested. Access to the data within the resource is facilitated by including legal information on authorship, rights and policies as well as technical information such as data resolution, media type and file size. Whereas a FAIR Data Point never contains the 'real' data, it can point to a public or secured download location. The FAIR Data Point (FDP) of the GENTURIS registry can be found at: https://genturis-registry.eu/api/fdp.

GENTURIS registry discovery

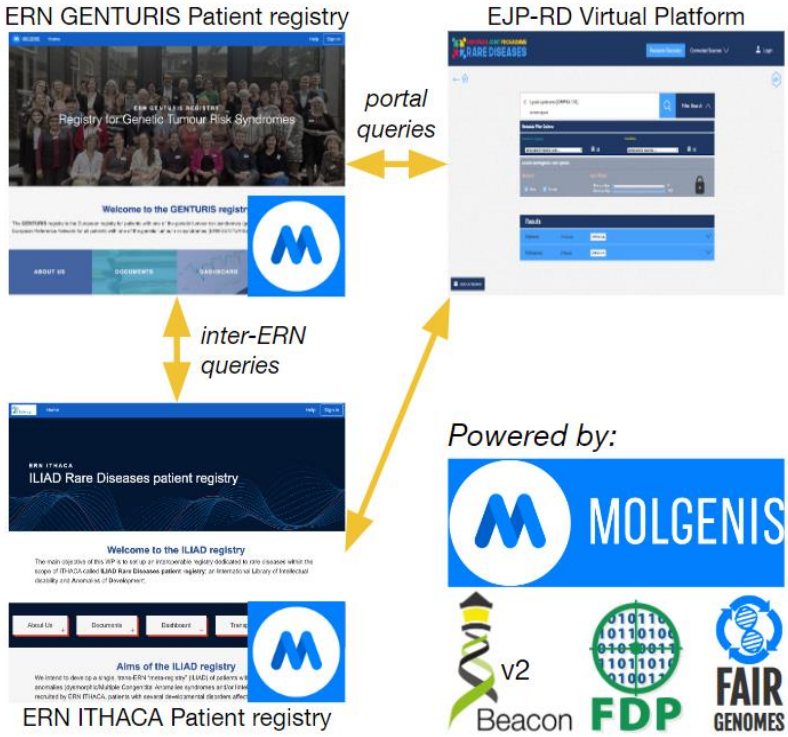
As researcher you may want to find registries and information in your field of interest. For instance, how many females diagnosed with Lynch syndrome at around age 40 are present in a certain registry? The registry may have no issues providing just the number of patients (e.g. 15), but wants to make absolutely sure that no further information about these 15 patients is accessible to others. To solve this problem, the Beacon system was created. It is intended to share non-sensitive information from a sensitive source and captures data for concepts such as cohorts, subjects, biosamples and genetic variation. Beacon is a Global Alliance for Genomics and Health (GA4GH) approved standard to exchange data at different levels of detail, including record-level (all information), count (only the total number) and boolean (yes or no).

Beacon can act as a bridge between systems, for instance, with the EJP-RD Virtual Platform. The Virtual Platform is a central search portal developed within the European Joint Programme on Rare Diseases. It lets people search for information about rare diseases in many different databases, registries, biobanks and knowledge bases all at once. Beacon is used to connect these resources to the central (discovery) index of the Virtual Platform. In the platform, people can use filters to search for specific characteristics, like gender, age and country. More powerful query options will only be available to authorized users.

The latest MOLGENIS version includes Beacon v2 to allow secure cross-ERN data queries, and this functionality is available within the GENTURIS registry. In the future the GENTURIS registry will be one of the connected sources that can be discovered though the Virtual Platform. The Virtual Platform is available at https://vp.ejprarediseases.org.

Showcase connecting registries of ERNs GENTURIS and ITHACA

An ERN may want to know if patients matching certain criteria have been registered at other ERNs as well. For instance, patients with PTEN Hamartoma Tumour Syndrome (PHTS) are included in ERN GENTURIS and ERN ITHACA (Intellectual disability, TeleHealth, Autism and Congenital Anomalies).
As a proof-of-concept, ERN GENTURIS is currently working with ERN ITHACA to make it possible to ask relevant questions to each other's registries without disclosing any sensitive information. Like the GENTURIS registry, the Iliad registry of ERN ITHACA is also making use of the MOLGENIS platform and is
therefore suitable for connecting registries to answer common questions that are relevant to both ERNs. For instance, how many patients are there with a molecular diagnosis that involve the *PTEN* gene (causing PHTS)? The Beacon implementation has been expanded to support such questions. These new queries can be combined with existing filters on sex, age, diagnosis, and so on. At the moment, the technical connections are in place, but these are intended to be used solely by computers. For humans, a user-friendly web pages will be developed where these questions can be created, send out, and the answer shown to the user.



Patient portal

Besides the GENTURIS registry 'professional portal' in which centres and health care professional can participate, a GENTURIS registry patient portal is currently under development. The patient portal will include functionalities for active participation by patients in the GENTURIS registry. The first release of the patient portal is expected soon.
Stay tuned for more information about the patient portal after the Summer break.

More information:

For more information about the GENTURIS registry,
visit https://genturis-registry.eu/

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