

What's new this month?

May 2021

EJP RD HIGHLIGHTS

APRIL 30TH

EJP RD Technical Hands-on Workshops: GA4GH Phenopackets

In view of the European Reference Networks (ERNs) Conference organized by the EJP RD on January 15th, ERN Technical Hands-on Workshops are being organized. The primary target audience is ERN Registry Developers/Programmers or Data Stewards for the practical aspects of the FAIRification tasks.

This two-hour workshop on **April 30th** provides an overview of the GA4GH Phenopackets standard, its applications, and the adoption benefits, specifically for ERNs.

The workshop topics include:

- Introduction to GA4GH, Phenopackets and adoption benefits
- Phenopackets schema (v1 and introduction to v2)
- Top-level elements of Phenopackets
- Use of Phenopackets
- EJP RD semantic Phenopackets



[More information](#)

MAY 27TH

Resource Webinar: DECIPHER



As part of the [EJP RD Resource Webinar series](#), the next webinar will be dedicated to [DECIPHER](#) and will be held on **May 27th**. DECIPHER is an online data-sharing platform facilitating the visualization and contextualization of rare disease genomic and phenotypic relationships, to enable diagnosis and discovery. Participants will be given an overview of DECIPHER, including the type of

patient data deposited in DECIPHER, and DECIPHER's data sharing model. They will also learn how to find relevant patient records and be introduced to the variant interpretation interfaces within the platform.

[More information](#)

ONGOING

MOOC on Rare Diseases Research Topics

Coordinated by the [Foundation for Rare Diseases](#), the EJP RD has launched its first **100% online academic education course (MOOC)** on rare diseases research topics, specifically addressing research issues related to diagnosis.

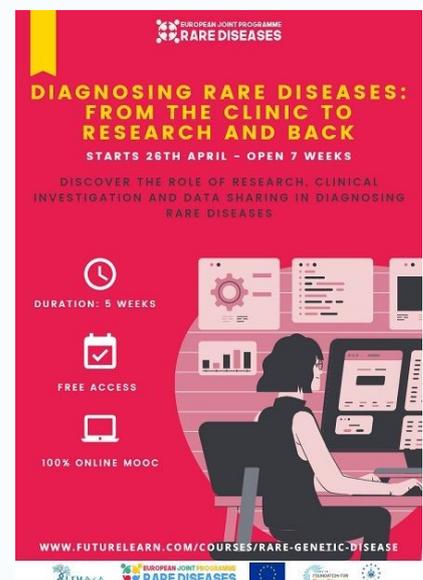
The MOOC is titled "**Diagnosing Rare Diseases: from the Clinic to Research and Back**"

Duration: 5 weeks (open for 7 weeks from April 26th)

Weekly study: 3 hours

Who is the course for?

While primarily designed for medical students and PhD/post-doc students in biomedical sciences, it will also be of interest to Patients Advocacy Organizations' representatives, healthcare professionals or paramedics who want to further their knowledge of rare diseases diagnosis.



[More information](#)

FORTHCOMING

ERN Workshop: Advisory Committee for Therapeutics (ACT)

Registration deadline: May 10th



In the context of the EJP RD [ERN Research Training Workshop](#) funding opportunity, an **Advisory Committee for Therapeutics (ACT) workshop** will be organized on **July 7th–8th**. The main objective of this workshop is to demonstrate the benefits of adopting the ACT model in rare disease communities, oriented around the **European Reference Network** groupings (and sub-domains), and to communicate best practices in this respect from the neuromuscular field. Participants will be given dedicated space and time to discuss and explore the feasibility of creating an ACT within their own disease domain and/or between disease domains.

[More information](#)

EJP RD FUNDING OPPORTUNITIES

Next collection date: June 1st

Networking Support Scheme (NSS) Funding Opportunity

Next collection date: June 1st

The aim of the NSS call is **to encourage knowledge-sharing between health care professionals, researchers and patients** on rare diseases and rare cancers, as well as **to enable or increase the participation of usually underrepresented countries in Europe in new and existing research networks**. Eligible applicants are **health care professionals, researchers, and patient advocacy organizations** from the following countries involved in the EJP RD: Armenia, Austria, Belgium, Bulgaria, Croatia, Czech Republic, Denmark, Estonia, Finland, France, Germany, Georgia, Greece, Hungary, Ireland, Israel, Italy, Latvia, Lithuania, Luxembourg, Malta, Norway, Poland, Portugal, Romania, Serbia, Slovakia, Slovenia, Spain, Sweden, Switzerland, the Netherlands, Turkey, United Kingdom. There is no limit on the number of participants per event; however, the maximum budget that can be requested is **€ 30,000 per networking event**. **The next collection date is June 1st at 14:00 (CET)**. To get more information and to apply, click below.

[More information](#)

[All EJP RD open funding opportunities here](#)



EJP RD IN EVENTS

During the month of April, EJP RD was presented at the following events:

- During a **Friends of Europe** event titled ***A European health data landscape for rare diseases (April 21st) at the round table***, where experts and policymakers gathered for a workshop on the future of Healthcare Digitalization. Focusing on rare diseases, they discussed the need for an EU-wide Health Data Space. Read more about this event [here](#).

NEWS FROM THE INTERNATIONAL RARE DISEASES RESEARCH CONSORTIUM (IRDiRC)

Leadership and Membership Changes

IRDiRC thanks **Dr. Sarah Bowdin** and **Mrs. Yukiko Nishimura** for serving as **Vice Chair** of the **Diagnostics Scientific Committee** and the **Patient Advocates Constituent Committee**, respectively.

IRDiRC warmly welcomes **Dr. Anneline Jonker**, who was elected **Vice Chair** of the **Therapies Scientific Committee**.

[More information](#)

22nd Meeting of the IRDiRC Consortium Assembly and Scientific Committees

IRDiRC **members** and **experts** from all over the world gathered on **March 15th** to the IRDiRC Consortium Assembly and Scientific Committees. The goal of the meeting was to provide the members with an update on ongoing activities and **present the 2021 roadmap of activities**. To see the available **Consortium Assembly meeting reports** (up to 2020), please click on the link below.

[More information](#)

RARE Conversations Webinar on Research in Rare Diseases: Specificities and Needs

Alexion's Rare Conversations invites you for its third webinar (Take Three) on Research in Rare Diseases: Specificities and Needs.

The webinar will take place on **May 5, 2021 from 14.00 – 15.30 CET.**

The event, in cooperation with BPI, **is aimed at discussing how the revision of the OMP Regulation (N° 141/20000), 20 years after its adoption, could trigger new research for the benefit of rare disease patients.**

[More information](#)

EuropaBio Advanced Therapies Webinar on Value of Advanced Therapies

EuropaBio invites you for its first webinar in the Advanced Therapies series of events, interviews, and publications titled "Taking health up a gear: Value of advanced therapies."

The webinar will take place on **May 11, 2021 from 12.00 – 13.00 CET.**

The event will discuss how **innovation in gene and cell-based medicine can bring a transformative value for patients**, with themes that include the cutting-edge science behind gene therapies, innovation in healthcare for cancer patients, and the role of EU policies in harnessing the potential of advanced therapies.

[More information](#)

European Conference on the Diffusion of Genomic Medicine: Health Economics & Policy

As part of **Solve-RD** (a European H2020 project on genetic research), the Health Economics Team of the Economics Laboratory of Dijon (University of Burgundy) is **organizing the European Conference on the Diffusion of Genomic Medicine: Health Economics & Policy** from **May 26th to 28th, 2021.**

To get more information and to **register (before May 24th)**, click below.

[More information](#)

CAREERS

Job opportunities are available at EJP RD and its member institutions:

- The EJP RD Coordination Team is looking for a **Junior Project Manager**
- Institute of Human Genetics, University Hospital Heidelberg is offering a **post-doc position**
- EATRIS is looking for a **Data Director**
- ERN-EYE, Hôpitaux Universitaires de Strasbourg, France is looking for a **Scientific Project Manager**



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