

European Joint Programme on Rare Diseases

Online Training on Biobanks for Rare
Diseases

Toward Innovative Research Biobanks for Rare Diseases: Overcoming the Challenges

29-30 October 2020

Online Training

PRELIMINARY PROGRAM

As part of the work in EJP RD Pillar 3: Capacity Building & Empowerment

Organised by partner ISCIII (Madrid, Spain)



This Training has received funding from the European Union's Horizon 2020 Research and Innovation Programme under Grant Agreement No 825575 - European Joint Programme on Rare Diseases

SUMMARY ABOUT THE TRAINING

This Training is aimed at **biomedical researchers, medical professionals and biobank managers** who want to approach **innovative biomedical research** projects on human biological samples especially focusing on **rare diseases**. We welcome the participation of representatives from patient organisations, as well. This Training covers one of the tasks planned in the EJP RD for the training on biobanks for rare diseases.

In two modules, several key issues in biomedical research involving human subjects, human biological samples and associated medical data will be addressed.

In the **first module** the role of the rare diseases biobanks in research will be put in place, considering the biobanking activity in the context of the European Reference Networks (ERNs), the unmet needs, the key role in unsolvable cases with rare diseases and, through specific biobanks' participation, this Training will provide excellent experiences and practical issues on the activity, for instance, regarding biobanks for brain tissues, myoblastoid cell lines and other.

In the **second module**, innovative considerations will be presented regarding the Human pluripotent stem cell registry, or patient-derived organoids applied to treatment research, and in general biobanking on advanced clinical materials for promoting clinical research on rare diseases. In a second part of this module, the governance of cell line biobank, the patient's perspective on biobanking for rare diseases research, and ethical and legal issues related to this activity will be prompted for further discussion after all the sessions of the Training, giving space to all the attendees for such dialogue.



GENERAL INFORMATION

Due to the worldwide situation regarding COVID-19, the international travel policies and extraordinary measures to limit the spread of SARS-Cov-2 virus, it seems reasonable to adopt a precaution approach by avoiding trips and face-to-face events until a safer scenario will arrive. In order to ensure the correct progress of the planned tasks of the [European Joint Programme on Rare Diseases \(EJP RD\)](#), [ISCIII \(Instituto de Salud Carlos III, of Spain\)](#) and the EJP RD leaders ensure that the Training on Biobanks for Rare Diseases, that was planned to be held in ISCIII, Madrid (Spain) on October 29-30, 2020, **will be held ONLINE**.

RELEVANCE, INTRODUCTION AND OBJECTIVES

The “**Training on Biobanks for Rare Diseases - Toward Innovative Research Biobanks for Rare Diseases: Overcoming the Challenges**” is part of a series of training activities proposed by the EJP RD. EJP RD is a European Commission funded project (grant agreement No 825575, 2019–2023) with the goal “to create a comprehensive, sustainable ecosystem allowing a virtuous circle between research, care and medical innovation”. For more information about the EJP RD, see <https://www.ejprarediseases.org/>

In particular, this Training on Biobanks for Rare Diseases (RD) is a part of the Work Package 14 of the EJP RD, focused on “Data Management & Quality Training”, and which aims to organize residential training courses in different countries.

The Training is organized by [Instituto de Salud Carlos III \(ISCIII\)](#) of Spain, in close collaboration with, mainly, EJP-RD task leader [Fondazione Telethon \(FTELE\)](#) of Italy. Other Project partners in this training series are BBMRI-ERIC, Charité – Universitätsmedizin Berlin, Hacettepe University Turkey, Medical University Gdansk Poland, University Medical Centre Groningen the Netherlands and Vilnius University Hospital Lithuania.

The Institute for Rare Diseases Research (IIER) of ISCIII runs the [Spanish National Biobank on Rare Diseases \(BioNER\)](#) and has a long experience in the field. In fact, its Director and staff currently hold the Scientific Coordination and Secretariat of the [EuroBioBank](#) network of RD biobanks. Also, IIER has a vast experience by organizing numerous courses focused on different aspects of rare diseases.

Biological samples from RD patients are precious resources for research. Due to their intrinsic rarity, these samples are often dispersed in multiple biobanks and are difficult to locate. The accessibility of these biological samples underpins the development of new diagnostic techniques, biomarker development, identification of potential therapeutic targets and testing therapeutic response.



Biobanks are key research infrastructures that offer professional services to the research community for sample sourcing and management. They play an important role in maintaining the quality of the biological samples from time-of-collection to distribution, as well as conserving the privacy and wishes of the patients. Biobanks have begun to share RD sample datasets to centralised sample catalogues (eg. [RD-Connect](#), [EuroBioBank](#)) to facilitate sample access and support the change towards best practice with their special position interfacing patients, clinicians and researchers. However, RD samples and associated data pose particular management challenges for their high variety of data types (e.g. disease names, genetic variants, phenotypes, clinical data), as well as balancing the need to share while preserving privacy and rights of RD patients.

The goal of this training series is to develop the capacity on data management of biobanks, allowing them to optimise operations to support the need of RD research community. Through the trainings, we aim to promote resources to be FAIR (Findable, Accessible, Interoperable and Reusable) and research reproducibility.

The training workshops will offer biobanks to learn how to harmonise and share their biological sample data encouraging direct interactions between RD clinicians/researchers and biobanks, and exchange on how to leverage biobanks in their RD research

The **general objective** of this training is to provide the trainees with knowledge, based on the experience of the expert speakers, on some innovative approaches which are currently relevant for the research on RD based on biobanks, tackling different challenges and their overcoming.

SPECIFIC LEARNING OBJECTIVES

Biobanks are key resources in order to increase research on rare diseases, timely and accurate diagnosis, tailor treatments, facilitate clinical trials, and support healthcare planning.

This training is composed of two training modules:

- In the **first module** (29 October 2020), the role of the rare diseases biobanks in research will be put in place, considering the biobanking activity in the context of the [European Reference Networks \(ERNs\)](#), the unmet needs, the key role in unsolvable cases with rare diseases and, through specific biobanks' participation, this Training will provide excellent experiences and practical issues on the activity, for instance, regarding biobanks for brain tissues, myoblastoid cell lines and other.
- In the **second module** (30 October 2020), innovative considerations will be presented regarding the human pluripotent stem cell registry, or patient-derived organoids applied to treatment research, and in general biobanking on advanced clinical materials for promoting clinical research on rare diseases. In a second part of this module, the governance of cell line biobank, the patient's perspective on biobanking for rare diseases research, and ethical and legal issues



related to this activity will be prompted for further discussion after all the sessions of the Training, giving space to all the attendees for such dialogue.

LEARNING METHOD

In an on-line format, there will be plenary presentations and dedicated interactive question & answers moderated sessions between speakers and participants, giving space for discussion to all.

PARTICIPANTS AND REGISTRATION

The training course is open to the international research community, clinicians, medical specialists, RD biobank's managers, healthcare professionals and RD patients' representatives.

To ensure active participation and exchange with teaching staff and participants, a maximum of 30 attendees will be admitted to each Training module. A selection process will be applied by an ad-hoc committee (composed by the EJP RD representatives who organized the Training), using as selection criteria the participants' background, their role with reference to rare diseases biobank activities, especially those linked to research, participation in ERNs or their involvement in the national plans for rare diseases in their country.

REGISTRATION

Online registration form is available at: [Registration link](#), until 28 September 2020.

An e-mail will be sent, by 9 October 2020, to the selected participants for the course. Respondents who are not contacted by email should consider themselves not selected but will be kept on a waiting list until 16 October 2020.

FEES AND COSTS

The training and registration is free of charge.
The training organisers will not cover expenses incurred by the participants in any case.

LEARNING ASSESSMENT AND IMPACT:

At the end of the Training an online questionnaire can be addressed to participants for learning and impact assessment.



ATTENDANCE CERTIFICATES

At the end of the course a certificate of attendance will be handed to the participants who attended 100% of the single training module or the entire Training program. No credits of Continuing Education in Medicine will be issued.

OFFICIAL LANGUAGE

English

VENUE

Online. Details for the connection will be provided in advance to confirmed participants.

CONTACT

If you have questions, please contact the course organisers through this email address: eva.bermejo@ejprd-project.eu

Please, indicate in the Subject: Training on Biobanking for RD



TOWARD INNOVATIVE RESEARCH BIOBANKS FOR RARE DISEASES: OVERCOMING THE CHALLENGES

Preliminary Program

Thursday, 29 October 2020 (Module 1)

10:45 – 11:00 Checking the online system, connection and sound testing for participants

Opening Session. Rare Diseases Biobanks and Research

Chair/Moderator: Mary Wang (FTELE, EJP RD, Italy)

- 11:00 – 11:15 Welcome address; Overview of the European Joint Program on Rare Diseases (EJP RD)
Eva Bermejo (ISCIII, EJP RD, Spain),
- 11:15 – 11:30 Welcome address; Role of rare diseases biobanks in the biomedical research, goals of the Training
Manuel Posada (ISCIII, EBB, EJP RD, Spain)
- 11:30 – 11:50 Samples in the context of the ERNs. The experience of a rare diseases biobank working in a big hospital participating in ten different ERNs.
Isabel Novoa (Vall d'Hebron Hospital's Biobank, Spain)
- 11:50 – 12:10 Rare diseases biological samples and unmet needs
Alex Felice (Umalta, EBB, BBMRI-MT, Malta)
- 12:10 – 12:30 Undiagnosed Rare Diseases International (UDNI): Type of biological samples used for solving unsolvable cases
Estrella López (ISCIII, EJP RD, Spain)
- 12:30 – 13:00 Moderated discussion – Questions and answers
- 13:00 – 15:00 Break

Rare Diseases biobanks and research outcomes

Chair/Moderator: TBC

- 15:00 – 15:15 EJP RD: Overview of its tools related to biological samples
Mary Wang (FTELE, EJP RD, Italy)
- 15:15 – 15:35 Brain tissues biobanks and rare diseases
Alberto Rábano (CIEN Network, Spain)
- 15:35 – 15:55 Lymphocyte Cell Lines biobank and rare diseases
TBC
- 16:00 – 16:30 Moderated discussion I – Questions and answers
- 16:30– 16:50 Myoblastoid cell lines
Hanns Lochmüller (University of Ottawa, Canada)
- 16:50 – 17:00 Moderated discussion II – Questions and answers
- 17:00 End of Module 1 – End of Day 1



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Friday, 30 October 2020 (Module 2)

Biobanking on advanced clinical materials for promoting clinical research on rare diseases

Chair/Moderator: Manuel Posada (*ISCIII, EBB, EJP RD, Spain*)

- 11:00 – 11:05 Recap of Day 1 and aims of Day 2
Manuel Posada
- 11:05 – 11:25 The Human pluripotent stem cell registry
Nancy Mah (EJP RD, Charité Universitätsmedizin Berlin, Germany)
- 11:25 – 11:50 Clinical trials and biobanking
TBC
- 11:50 – 12:10 Curing genetic disease in patient-derived organoids / Patient-derived organoids applied to treatment research
TBC
- 12:10 – 12:30 Organoids: a patient in the lab; Organoids and rare diseases biobanks
Beatriz Martínez (ISCIII, RD National Biobank - BioNER, Spain)
- 12:30 – 13:00 Moderated discussion – Questions and answers
- 13:00 – 15:00 Break

Biobanking for rare diseases, patients and ELSI issues

Chair/Moderator: TBC

- 15:00 – 15:20 Governance of a cell line biobank
TBC
- 15:20 – 15:40 Biobanking on Rare Diseases: Patients' perspective.
Simona Borroni (Advisory Board TNGB, Board Dravet Europe, "Gruppo Famiglia Dravet", Italy)
- 15:40 – 16:00 Data collection and biobanks
TBC
- 16:00 – 16:20 Ethics and laws applied to innovative biological sample use in research
Michaela Th. Mayrhofer & Mónica Cano-Abadía (BBMRI-ERIC, Austria)
- 16:20 – 16:45 Moderated discussion – Questions and answers
- 16:00 – 17:00 End of Module 2 – End of Day 2 and Training

Closure



Confirmed List of Speakers & Chairs

Eva Bermejo	Instituto De Salud Carlos III, Spain; EJP RD
Simona Borroni	Dravet Europe, "Gruppo Famiglia Dravet", Italy
Mónica Cano-Abadía	BBMRI-ERIC, Austria; EJP RD
Alex Felice	Umalta, BBMRI-MT, Malta; EuroBioBank
Hanns Lochmüller	University of Ottawa, Canada
Estrella López	Instituto De Salud Carlos III, Spain; EuroBioBank; EJP RD
Nancy Mah	Charité Universitätsmedizin Berlin, Germany; EJP RD
Beatriz Martínez	Instituto De Salud Carlos III, RD National Biobank - BioNER, Spain
Michaela Th. Mayrhofer	BBMRI-ERIC, Austria; EJP RD
Isabel Novoa	Vall d'Hebron Hospital's Biobank, Spain
Manuel Posada	Instituto De Salud Carlos III, Spain; EuroBioBank; EJP RD
Alberto Rábano	CIEN Network, Spain
Mary Wang	Fondazione Telethon, Italy; EJP RD

