



TRAINING WORKSHOP

**ORGANIZING & MAXIMIZING RARE DISEASE
BIOLOGICAL SAMPLE DATA IN BIOBANKS**

1 – 2 April 2019, Milan

GENERAL INFORMATION

INTRODUCTION AND OBJECTIVES

This training workshop is a part of a series of capacity building programs put forth by the [European Joint Programme on Rare Diseases](#) (EJPRD). EJPRD 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”.

The organisation of this workshop series on biobanks and biological samples is coordinated by Fondazione Telethon, Italy. Project partners in this training series are BBMRI-ERIC, Charité – Universitätsmedizin Berlin, Institute of Health Carlos III Spain, Hacettepe University Turkey, Medical University Gdansk Poland, University Medical Centre Groningen the Netherlands and Vilnius University Hospital Lithuania.

Biological samples from rare disease (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 workshops, we aim to promote FAIR of resources and research reproducibility. The training workshops will offer biobanks to learn how to harmonise and share their rare disease biological sample data and encourage direct interactions between RD clinicians/researchers and biobanks, where researchers can exchange on how to leverage biobanks in their RD research.

2019 WORKSHOPS & THEME

Several training workshops on rare disease sample management will be organised during the EJPRD run time.

In 2019, the theme is “**Organizing & maximizing rare disease biological sample data in biobanks** “. Two workshops are available in 2019 and will have similar contents. The two workshops may vary only slightly to tailor the local organisers logistical or needs. Workshop themes may vary from year to year, based on feedback from participants and EJPRD project outputs.

Workshop 1 1-2 April 2019, Milan, Italy

Workshop 2 29-30 October, Vilnius, Lithuania

LEARNING METHOD

The training workshop will consist of plenary lectures and interactive small-group exercises, based to the *Problem-Based Learning* (PBL) method. PBL is a highly interactive and learner-centred approach where the learning occurs by working in a small group assisted by a facilitator to develop a solution of a problem. Practical demonstration of biobank or sample

catalogue related tools are also included. Participants are asked to bring their **laptops** in order to participate in the PBL and the practical demonstrations.

PARTICIPANTS AND REGISTRATION

The training workshop is open to healthcare professionals, researchers, clinicians, biobank managers, database managers and rare disease patient advocates. In particular, we welcome participants from all types of biobanks (academic, hospital, private) with focuses on rare diseases.

To ensure active participation and exchange with teaching staff and fellow participants, a maximum of 25 participants will be admitted to the training workshop on *first-come, first-served basis*. Up to 4 places are reserved for patient advocates or representatives.

Registration is open for Workshop 1 - Milan only.

Registration Form:

Travel Fellowships - Application deadline 4 March 2019: Two travel fellowships are available to cover one round trip flights and up to two nights of hotel accommodation, for a total of up to 470euros. We especially encourage participants from EU-13 countries, if you wish you apply for a travel fellowship please submit the completed registration with the fellowship section by 4 March 2019.

FEES AND COSTS

The workshop and registration is free of charge. Coffee refreshments and lunches will be offered during the workshop. Participants must arrange their own travel, accommodation and other expenses to attend the workshop. The workshop organisers will not cover expenses incurred by the participants.

ATTENDANCE CERTIFICATES

At the end of the workshop a certificate of attendance will be handed to the participants who attended the entire workshop. No credits of Continuing Education in Medicine will be issued.

LANGUAGE

Workshops will run in English

CONTACT

If you have questions please write to the workshop organiser Dr Mary Wang mwang@telethon.it

Preliminary Program of the Workshop

DAY 1 "What do I need to consider"

- 08:30 - 09:00 Participants registration
 09:00 - 09:30 Welcome address & Faculty
 Presentation of the course and EJP-RD, Mary Wang, Fondazione Telethon
 09:30 - 09:45 Introduction to Problem Based Learning, (Claudio Carta, Istituto Superiore di Sanità)
 09:45 - 11:00 *PROBLEM ANALYSIS* - Reading and analysis of the problem in groups with facilitators
 11:00 - 11:15 Break
 11:15 - 12:15 **Lecture 1** - Rare disease biobanking and network state-of-art, Manuel Posada, Institute of Health Carlos III
 12:15 - 12:45 **Lecture 2** - Principles of sample data sharing and MIABIS, David van Enckevort, University Medical Center Groningen
 12:45 - 13:30 Lunch
 13:30 - 14:00 **Lecture 3** - Privacy management & case study, Michaela Th. Mayrhofer, BBMRI-ERIC
 14:00 - 16:00 *PROBLEM SOLUTION* - Group discussion
 16:00 - 17:30 Group presentations & Feedback from the trainers

DAY 2 "Getting Practical on Data Management"

- 08:30 - 09:00 Arrival/set up
 09:00 Aims of the day
 09:00 - 10:00 **Lecture 4** - Orphanet Nomenclature and Orphanet Rare Disease ontology (ORDO), Houda Ali, Orphanet
 10:00 - 10:15 **Tool Demo** - RD-Connect Sample Catalogue, Mary Wang, Fondazione Telethon
 10:15 - 10:45 Break
 10:45 - 12:45 **Practical session** - Hands-on exercise on data harmonization, David van Enckevort, University Medical Center Groningen, Mariska Slofstra, University Medical Center Groningen
 12:45 - 13:30 Lunch
 13:30 - 14:15 **Keynote lecture** - RD biobanks and working group in BBMRI Italy, Luca Sangiorgi, RD Working Group, BBMRI.IT
 14:15 - 15:30 **Tool Demo** - BBMRI-ERIC Negotiator, TBC, David van Enckevort, University Medical Center Groningen
 15:00 - 15:30 Concluding remarks, gathering of feedbacks

SPEAKERS

Mary Wang, Fondazione Telethon, Italy
 Claudio Carta, Istituto Superiore di Sanità, Italy
 David van Enckevort, University Medical Center Groningen, the Netherlands
 Manuel Posada, Institute of Health Carlos III, Spain
 Michaela Th. Mayrhofer, BBMRI-ERIC, Austria
 Houda Ali, Orphanet, France
 Luca Sangiorgi, RD WG BBMRI.IT, Italy
 Mariska Slofstra, University Medical Center Groningen, the Netherlands

