

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

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

Organised by partner ISCIII (Instituto de Salud Carlos III, 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, reviewing some of their research outcomes. This Training will provide excellent experiences and practical issues on the activity, for instance, regarding biobanks for brain tissues, patient-derived organoids applied to treatment research, and primary human cell lines for drug and biomarker discovery (with the example of myotonic dystrophy).

In the **second module**, innovative considerations will be presented regarding the Human pluripotent stem cell registry, clinical trials and biobanking, or the application of lymphoblastoid cell lines for studying CNS disorders, and again organoids seen as a way to have a proxy of the patient in the lab, and in general biobanking on advanced clinical materials for promoting clinical research on rare diseases. In a second part of this module, the issues related to data collection in biobanks, together with the patient's perspective on biobanking for rare diseases research, and ethical and legal issues applied to innovative biological sample use in research will be prompted for further discussion after all the sessions of the Training, giving space to all the attendees for such dialogue with the speakers and moderators.

In **both modules** it is expected (and will be stimulated) a high interaction between the trainees and the trainers in order **to promote the importance of biobanking activities in rare diseases research**.



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 at the Institute of Health Carlos III (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 the 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, reviewing some of their research outcomes. This Training will provide excellent experiences and practical issues on the activity, for instance, regarding biobanks for brain tissues, patient-derived organoids applied to treatment research, and primary human cell lines for drug and biomarker discovery (with the example of myotonic dystrophy).
- In the **second module** (30 October 2020), innovative considerations will be presented regarding the Human pluripotent stem cell registry, clinical trials and



biobanking, or the application of lymphoblastoid cell lines for studying CNS disorders, and again organoids seen as a way to have a proxy of the patient in the lab, and in general biobanking on advanced clinical materials for promoting clinical research on rare diseases. In a second part of this module, the issues related to data collection in biobanks, together with the patient's perspective on biobanking for rare diseases research, and ethical and legal issues applied to innovative biological sample use in research will be prompted for further discussion after all the sessions of the Training, giving space to all the attendees for such dialogue with the speakers and moderators.

In **both modules** it is expected (and will be stimulated) a high interaction between the trainees and the trainers in order **to promote the importance of biobanking activities in rare diseases research.**

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 a 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

Thursday, 29 October 2020 (Module 1)

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

Session 1 - 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 Institut de Recerca (VHIR), 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

Session 2 - Rare Diseases biobanks and research outcomes

Chair/Moderator: Meral Özgüç (Hacettepe University, Turkey)

- 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 Foundation Brain Tissue Bank, Spain)
- 15:35 – 15:55 Patient-derived organoids applied to treatment research
Robert Vries (Hubrecht Organoid Technology, The Netherlands)
- 16:00 – 16:30 Moderated discussion I – Questions and answers
- 16:30– 16:50 Primary human cell lines for drug and biomarker discovery in myotonic dystrophy
Hanns Lochmüller (University of Ottawa, Canada; EJP RD: University of Freiburg, Medical Centre, Germany & CNAG/CRG, Barcelona, Spain)
- 16:50 – 17:00 Moderated discussion II – Questions and answers
- 17:00 End of Module 1 – End of Day 1



Friday, 30 October 2020 (Module 2)

Session 3 - 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/ Fraunhofer IBMT, Germany)
- 11:25 – 11:50 Clinical trials and biobanking
Rosita Kammler (ESBB, IBCSG, ETOP, Switzerland)
- 11:50 – 12:10 Applying lymphoblastoid cell lines for studying CNS disorders
David Gurwitz (Tel-Aviv University, Israel)
- 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

Session 4 - Biobanking for rare diseases, patients and ELSI issues

Chair/Moderator: Lorena Casareto (*Istituto Ortopedico Rizzoli, TNGB, EBB, Italy*)

- 15:00 – 15:20 Data collection and biobanks
Ma'n Zawati (McGill University, Center of Genomics & Policy, Canada)
- 15:20 – 15:40 Biobanking on Rare Diseases: Patients' perspective.
Simona Borroni (Advisory Board TNGB, Board Dravet Europe, Gruppo Famiglie Dravet, Italy; Dravet syndrome European Federation, France)
- 15:40 – 16:00 Ethics and laws applied to innovative biological sample use in research
Michaela Th. Mayrhofer & Mónica Cano-Abadía (BBMRI-ERIC, Austria)
- 16:00 – 16:30 Moderated discussion – Questions and answers
- 16:30 – 16:45 End of Module 2 – End of Day 2 and Training
Manuel Posada
- Closure



Confirmed List of Speakers & Chairs/Moderators (alphabetic order of surnames)

Eva Bermejo-Sánchez	Instituto De Salud Carlos III, Spain; EJP RD
Simona Borroni	Gruppo Famiglie Dravet, Italy; Dravet syndrome European Federation, France
Mónica Cano-Abadía	BBMRI-ERIC, Austria; EJP RD
Lorena Casareto	Istituto Ortopedico Rizzoli, Italy; Telethon Network of Genetic Biobanks; EuroBioBank
Alex Felice	UMalta, BBMRI-MT, Malta; EuroBioBank
David Gurwitz	Tel-Aviv University, Israel; EuroBioBank
Rosita Kammler	ESBB, IBCSG, ETOP, Switzerland
Hanns Lochmüller	University of Ottawa, Canada; University of Freiburg, Medical Centre, Germany & CNAG/CRG, Spain; EJP RD
Estrella López-Martín	Instituto De Salud Carlos III, Spain; EuroBioBank; EJP RD
Nancy Mah	Charité Universitätsmedizin Berlin/ Fraunhofer IBMT, Germany; EJP RD
Beatriz Martínez-Delgado	Instituto De Salud Carlos III, RD National Biobank - BioNER, Spain
Michaela Th. Mayrhofer	BBMRI-ERIC, Austria; EJP RD
Isabel Novoa	Vall d'Hebron Institut de Recerca (VHIR), Spain
Meral Özgüc	Hacettepe University, Turkey
Manuel Posada de la Paz	Instituto de Salud Carlos III, Spain; EuroBioBank; EJP RD
Alberto Rábano	CIEN Foundation Brain Tissue Bank, Spain
Robert Vries	Hubrecht Organoid Technology, The Netherlands
Mary Wang	Fondazione Telethon, Italy; EJP RD
Ma'n Zawati	McGill University, Center of Genomics & Policy, Canada



BIOSKECHTS OF THE SPEAKERS AND MODERATORS

Eva Bermejo-Sánchez



Tenured Scientist of ISCIII (Institute of Health Carlos III), Chief of Area at IIER (Institute for Rare Disease Research), and Scientific Director of the National Biobank of ISCIII. Head of the Research Unit on Congenital Anomalies (UIAC) of IIER. Scientific Coordinator of ECEMC (Spanish Collaborative Study of Congenital Malformations) and its Clinical Network, and Responsible for the Clinical Genetics and Epidemiology Section. Collaborator of SpainUDP, the Spanish Undiagnosed Rare Diseases Program, of IIER. Researcher of ICBDSR and EUROCAT. Principal Investigator for ISCIII at the European Joint Programme on Rare Diseases (EJP RD). Former President of the Executive Committee of ICBDSR (International Clearinghouse for Birth Defects Surveillance and Research). Member of the Advisory Committee of FEDER (Spanish Federation of Rare Diseases). Member of the Clinical Advisory Committee of RDMM (the European Rare Disease Models & Mechanisms Network), of the project "Solve-RD - solving the unsolved rare diseases". Three Scientific awards. Accredited in Human Genetics. 37 funded projects/142 articles in indexed scientific journals/ 14 books or book chapters/ participation in more than 150 academic or educational activities. Organization of more than 45 scientific national or international meetings. Participation in the inception and organization of the World Birth Defects Day, since 2015. Evaluator of the Spanish National Agency for Evaluation and Prospective (ANEP).

Simona Borroni



Simona Borroni has been active in the field of rare diseases, as a volunteer in non-profit organisations since 2009. She is one of the founders and Chair of the Gruppo Famiglie Dravet, Board member of the Italian Federation of Epilepsies and of the Dravet Syndrome European Federation, member of the Advisory Board of the Telethon Network of Genetic Biobanks. She is Expert patient trained by EUPATI IT and participated, as patient representative reviewer, in the ERA PerMed Joint Transnational Call for Proposals 2019. She has been Member, in 2017/2018, of the Scientific Advisory Board for the Italian Whitepaper of epilepsy. She is responsible for the management of different kinds of projects, at a local and EU level,



including: International open Calls to fund Dravet syndrome research projects, Surveys run at a European level, Congresses and families meeting organisation. In 2018/2019 she participated in BBMRI Italy ELSI working Groups.

Mónica Cano-Abadía



Mónica Cano Abadía is a moral and political philosopher. In 2014, she obtained with honors her PhD in Philosophical Studies from the University of Zaragoza. In addition to her MA in Philosophical Studies (2010), she holds a second MA in Secondary Education Teaching (2017), with a specialization in the pedagogy of ethics and political philosophy. She has been a postdoctoral fellow at the Center for Advanced Studies – South East Europe (University of Rijeka), and at the Section of Political Philosophy (University of Graz). Since 2011, she has worked as a lecturer at the Universities of Zaragoza and Graz, where she has taught several courses on different topics related to ethics and moral philosophy. She is currently an ELSI Services Officer at BBMRI-ERIC, where she is charge of

coordinating the ELSI Helpdesk Network and the ELSI Knowledge Base.

Lorena Casareto



Lorena Casareto is the Project Manager of national and European projects on rare diseases. She has over 10 years of hands on experience in rare disease biobank networking. In particular, she has deep insights in biobank data harmonisation, sample workflows and the integration of IT infrastructure with external tools. Within the network, she also oversees customer services, communications, organisation of events and the drafting of policies and ELSI documents. Lorena participates actively in national and European biobank networks, as well as European rare disease infrastructure projects, providing advice and shaping the development of future tools for sample data management.



Alex Felice



Professor Alex E. Felice M.D., Ph.D., F.M.C.Path (spec. Genetika) is a graduate of the Medical School of the University of Malta (MD 1971 & M.Phil. 1975) and the School of Graduate Studies of the Medical College of Georgia, Augusta GA, U.S.A. (Ph.D. 1981) He held academic positions on the Faculty at Augusta in both the School of Medicine and the School of Graduate Studies (Associate Professor; Cell & Molecular Biology & Pediatric Hematology) and on the Research Service of the Veterans' Administration Medical Center (Augusta, GA, U.S.A.) as Program Director in Hemoglobin Research (Molecular Hematology) He was member of the US South-Eastern Regional Genetics Group and the Sickle Cell Network of Clinics in the state of Georgia.

In 1992 he was appointed Professor (Biomedical Sciences) at the University of Malta in the School of Medicine, and Visiting Thalassaemia Consultant in the Ministry of Health. Here, he directed the establishment of the Thalassaemia and Molecular Genetics services and the development of a Molecular Biotechnology Research Program including Human / Medical Genomics, the Malta BioBank, and the membership in Eurobiobank.

His main research interest is in globin gene control and the genomics of rare blood disease. Professor Felice is an elected member of several international societies in science, haematology and human genetics as well as Foundation President of the Malta Chamber of Scientists and Chairman of the consortium "European Researchers' Night, Science in the City" the city being Valletta, his home town. He is Malta representative on the management committee and the assembly of member states of the BioBanking and BioMolecular Resource Research Infrastructure of the EU (BBMRI-ERIC) and sits on the board of the "Rare Disease Alliance, Malta".

David Gurwitz



Dr. David Gurwitz obtained his PhD from the Faculty of Life Sciences at Tel Aviv University (Israel) in 1986 and completed postgraduate studies at the University of California, Irvine in 1989. In 1995, following a second postdoc with a pharmaceutical company, he joined the Department of Human Molecular Genetics at the Faculty of Medicine at his alma mater, where he is now Associate Professor. His research is focused on discovery of genomic biomarkers for precision medicine of CNS disorders. David is editorial board member of several journals including Trends in Molecular Medicine, Genome Medicine, CNS Drugs, and npj Genomic Medicine.

Website: <http://www.sagol.tau.ac.il/en/faculty/gurwitz-david/>



Rosita Kammler



Rosita Kammler is President of the European, Middle Eastern and African Society for Biopreservation and Biobanking (ESBB). She had already served ESBB as Vice-President for two years, and before that as Councilor.

Rosita Kammler heads the Translational Research Coordination at the International Breast Cancer Study Group (IBCSG) and the European Thoracic Oncology Platform (ETOP). Her work has revolved around clinical trial biobanking, translational research coordination and project management for the past 18 years. She has provided expertise and leadership for designing, building and conducting the translational research programs and biobanks for IBCSG and ETOP, enabling state-of-the-art oncology research. She is

responsible for ensuring that trial biosamples and associated data meet the objectives for quality, quantity, ethical use, to ensure the integrity of research data and analysis. These biobanks have been a critical resource for the more than 50 translational research studies conducted by IBCSG/ETOP.

Hanns Lochmüller



Hanns Lochmüller, Senior Scientist, CHEO Research Institute Professor of Neurology, University of Ottawa Faculty of Medicine and The Ottawa Hospital Department of Medicine.

Hanns is a neurologist and clinical academic specializing in genetic neuromuscular disorders and rare disease. He is Senior Scientist at the Children's Hospital of Eastern Ontario (CHEO) Research Institute. He also holds appointments as Professor of Neurology in the University of Ottawa Faculty of Medicine and the Department of Medicine, Division of Neurology at The Ottawa Hospital. He is affiliated with the University of Ottawa Brain and Mind Research Institute and

Department of Cellular and Molecular Medicine and with the Ottawa Centre for Neuromuscular Disease.

Hanns trained as a neurologist in Munich, Germany and in Montreal, Canada. From 2007 to 2017, he held the chair of experimental myology at the Institute of Genetic Medicine at Newcastle University in the UK. He continues to hold a scientific appointment at the Department of Neuropediatrics and Muscle Disorders of the Medical Center – University of Freiburg in Germany and as visiting scientist at the Centro Nacional de Análisis Genómico (CNAG), Centre for Genomic Regulation, Barcelona in Spain.

His research interests include molecular therapies of neuromuscular disorders; molecular pathogenesis of muscle and neuromuscular junction disorders; neurogenetics and translational research; data sharing and -omics in neuromuscular and rare diseases; and genomics and systems medicine. In addition to his scientific and clinical research interests, he is internationally



active in rare disease science policy and research collaborations. He chaired the Interdisciplinary Scientific Committee of the International Rare Diseases Research Consortium (IRDiRC) and the Executive Committee of the TREAT-NMD Alliance. He initiated and coordinated the highly successful "RD-Connect" international infrastructure for rare disease data and biosample sharing and analysis, is co-founder and former coordinator of the German muscular dystrophy network (MD-NET), and former scientific coordinator of EuroBioBank, a European (and Canadian) network of biobanks for rare disorders.

Hanns's clinical activities focus on clinical research and care of patients with rare neuromuscular disorders, including myotonic dystrophy (DM1), spinal muscular atrophy (SMA), muscular dystrophy and congenital myasthenic syndromes (CMS). He has a strong commitment to working with patients and patient organizations in Canada, as he has with organizations in Europe for many years.

Estrella López-Martín



Estrella López Martín, has a PhD in Pharmacy. Skills in Pharmacy, Biochemistry, Microbiology, Environmental Health and Rare Diseases research. Currently she has a Specialized Technician permanent position in IIER of ISCIII, participating in several tasks related to the Spanish Undiagnosed Rare Diseases Program SpainUDP. Also involved in activities derived from the European projects RD-Connect and Solve-RD, focused on infrastructures and research on RD. Expertise within the rare diseases field: interoperability strategies at international level, undiagnosed diseases programs, phenotype ontologies and research projects. Secretariat in EuroBiobank (EBB).

Nancy Mah



Nancy Mah (Human Pluripotent Stem Cell Registry, hPSCreg) has a background in molecular biology with a PhD in the field of Cell Biology, complemented with bioinformatics, and she has been working in the stem cell field for over a decade. Since joining the hPSCreg project in 2014, she has contributed to the project's data structure and management, which entails not only biological cell line data but also the ethical provenance of these lines. Her goals within the EJP-RD project are to make the hPSCreg interoperable with other resources in the Virtual Platform and to help educate others on FAIR data management through the biobank training workshops offered by EJP-RD.



Beatriz Martínez-Delgado



Beatriz Martínez is Senior Scientist at the Institute for Rare Diseases Research (IIER, ISCIII) and Group Leader of the Molecular Genetics Unit. She is involved in different research projects about genetic basis of different rare diseases, participates in the Undiagnosed Diseases Program (SpainUDP) of the IIER and in the Rare Diseases Biobank (BioNER) activities. She also collaborates in the Scientific Committee of the Spanish Registry of Patients with Alpha-1 Antitrypsin Deficiency (REDAAT), in the RD Committee of the Spanish Association of Human Genetics (AEGH), in the Advisory Committee of the Spanish Federation of Rare Diseases FEDER, as well as in the U-758 Group of the Center for Biomedical Research in Network Rare Diseases CIBERER.

Michaela Th. Mayrhofer



Michaela Th. Mayrhofer is a political scientist and historian by training. She was educated in Vienna, Louvain-la-Neuve, Essex and Paris. In 2010, she has earned her PhD from both the Ecole des Hautes Etudes en Sciences Sociales and the University of Vienna, which was shortlisted by the Austrian Society for Political Science for 'best thesis 2010'. Prior to her involvement in BBMRI-ERIC, she was investigator in several national and international research projects focusing on the politics of biotechnology and the life sciences, especially the governance of biobanks. Her academic career led to various positions in Austria, the UK and Switzerland. Since 2019, she serves as the Head of ELSI Services & Research of BBMRI-ERIC, was co-Interim Director (Feb-Aug 2020) and coordinates the Code of Conduct for Health Research initiative.



Isabel Novoa



Isabel Novoa helps biomedical researchers get the best work in the biomarker discovery field to improve healthcare. I do it as the Director of a Biobank Facility that provides expert processing services of patient-derived samples.

She got a PhD in Biochemistry and Molecular Biology by the Universidad Autónoma de Madrid, Spain. Since 2010 she joined the biomedical research center Vall d'Hebron Research Institute as a Biobank facility coordinator, and since 2013 she is the Biobank Director. Her Hospital University Vall d'Hebron Biobank is member of the Spanish Biobank Network since 2010.

Meral Özgüç



Meral Özgüç is Professor (Emeritus) of Medical Biology at the Faculty of Medicine, Hacettepe University, Ankara, Turkey where she received her PhD in Medical Biology in 1985. She completed her undergraduate studies at Fairleigh Dickinson University in chemistry and received masters degree in biochemistry from Columbia University in the USA. Immediately afterwards, she worked as a Research Associate in the Department of Research Hematology, Childrens Hospital of Philadelphia before joining Hacettepe Faculty in 1980. Besides research and teaching activities she served as Assistant Dean of the Faculty of Medicine (2006-2009) and Chair of the Basic

Medical Sciences Division (2010-2012.) She has founded the Hacettepe DNA/Cell Bank for Rare Diseases in 1995 and acted as scientific director until 2020. The biobank is a member of Eurobiobank since 2014.

Her work, supported by State Planning Agency, Scientific and Technical Research Council of Turkey (TÜBİTAK) and through international grants, focused on a genomic medicine approach for the study of rare diseases. She was actively involved in the formulation of national policies to create an awareness of and to promote genomics in public health. Her scientific publications concentrate in the area of identification of new disease genes and genomic variants and development of diagnostic tests. She was and still is involved in networking activities, governance and bioethical aspects of sample acquisition and data management of biobanks.

She has worked as a member of various international committees involved in genomics and health such as OECD - Working Party on Biotechnology and Human Health Related Biotechnologies, ESF - Integrated Approaches to Functional Genomics, EC-FP6 Genomics and Biotechnology for Health (National Contact Point), and UNESCO-International Bioethics Committee and National EPMA Board in the section for Neonatal Diagnostics and Population Screening. Currently she is a participant (HUGEN) of EJP-RD Pillar 3.



Manuel Posada de la Paz



Manuel Posada, Research Full Professor, MD, PhD in Medicine. Specialist in Internal Medicine and also specialist in Public Health and Preventive Medicine, Autonomous University, Madrid, Spain. Expert in areas such as multivariate analyses, medical statistics and research methods.

Current position: Director of the Institute of Rare Diseases Research (IIER), Institute of Health Carlos III, and lead of a broad range of rare diseases activities in Spain in areas such as epidemiological and public health research. He is also the Director of the National Biobank on Rare Diseases (ISCIII) and the National Rare Diseases Registry.

Director of a WHO-Collaborating Center 1996-2011.

He was acting as the Spanish representative (MoH) at the EC Action Plan on Rare Diseases (1999-2003) and after that in the EC Rare Diseases Task Force (2004-2009). Independent expert of the Commission Expert Group of Rare Diseases (CEGRD), European Commission and also of the Advisory Board of the European Commission Platform Rare Diseases Registration.

Also member of the Institutional Committee of the Spanish National Strategy of Rare Diseases on behalf the ISCIII and the Funders of Constitutional Committee of the International Rare Diseases Research Consortium (IRDIRC)

He was elected as a Secretary of ICORD (2012-2014), President-Elect (2014-2016) and President 2016-2019.

Project leader of several research projects and author of more than 150 papers in national and international peer review journals. Editor of the book entitled "Rare Diseases Epidemiology", Springer, 2010, and "Rare Diseases Epidemiology: Update", Springer, 2017

Alberto Rábano



Alberto Rábano, Dr med. Dr phil. – Spanish neuropathologist and basic neuroscientist, working since 2009 at the CIEN Foundation (Research Institute for Neurological Diseases), attached to the Carlos III Health Institute, as Head of Neuropathology and Scientific Director of the CIEN Tissue Bank. Academic education at the Complutense University of Madrid (MD), residency of Pathology at the Gregorio Marañón University Hospital in Madrid, and a fellowship in Pathology and Immunohistochemistry at Hammersmith Hospital, London, UK. PhD in Sciences with a dissertation on the comparative pathology of Alzheimer's and argyrophilic grain tauopathies.

Previously to his current position, extensive work as Head of Pathology and Research Director of a University Hospital in Madrid (Fundación Hospital Alcorcón), taking part during this period (1997-2009) and thereafter in the development of four brain banks in different cities (Madrid, Murcia, Salamanca and Córdoba). During the "mad cow" crisis (2000-2009) he was in charge of the neuropathological diagnosis of prion diseases in



several regions of Spain, including Madrid, and was a member of several governmental committees dealing with prion disease surveillance and biosafety issues. During this period he took part in the neuropathological diagnosis of 4 of the 5 cases of variant Creutzfeldt-Jakob disease identified in Spain.

He has participated in several research ethical committees including the Ethical Advisory Board of the Human Brain Project (European Union) (2014 – 2018) and of the Steering Committee of the Spanish National Biobank Network (2014 – 2020), and is the current President of the Spanish Neuropathology Club. He has received several media and scientific awards for the promotion of brain donation and for research in Alzheimer's disease (Spanish Society of Neurology Award, 2017).

His research interests are focused on diagnostic and molecular neuropathology of neurodegenerative diseases, biospecimen science and biobanking, and ethical issues related to tissue donation for research. Since 2014 he is principal investigator of the core research program at the Alzheimer's Center Queen Sofía Foundation, in Madrid, based on a cohort of over 450 institutionalized patients with moderate to advanced dementia with periodic follow-up, biochemistry, MR and brain donation. In recent years, he took an active part in the research line that led to the discovery of the presence of brain fungal colonization in several neurodegenerative diseases and in breakthrough research in adult hippocampal neurogenesis. He organized the 1st Symposium on Brain Banks (Salamanca, Spain, September 2018), the first National meeting of brain banks in Spain. He is author or co-author of 136 original scientific papers, h-index 36, and 6 book chapters.

Robert Vries



Robert Vries, CEO, Hubrecht Organoid Technology (HUB)

Robert received his PhD in Biochemistry from the Leiden University Medical Center on a molecular study of oncogenic transformation. He subsequently moved to Stanford University (USA) to do his Post Doc studying neural stem cells. Upon his return to the Netherlands he continued the study of adult stem cells in the group of Prof Hans Clevers at the Hubrecht Institute in The Netherlands.

In the group of Hans Clevers he was part of the team that developed the breakthrough technology that allowed the expansion of adult stem cells. The so-called

Organoid Technology became the basis of the non-profit company 'Hubrecht Organoid Technology' (HUB) of which he is currently the CEO.

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Mary Wang



Chihui Mary Wang is a Partnership & Project Manager at Fondazione Telethon. Mary has over 15 years of professional experience in biomedical research and has been contributing and leading in numerous European initiatives related to research policy and data infrastructures. Currently, Mary is involved in the EJP RD as task leader on training, focusing on optimization of biological sample data management and harmonization; responsible for delivery of training program and 10 training courses. In addition, she co-chairs the Use Cases Work Focus in EJP RD and is responsible for capturing researcher needs as the base for co-design of the federated Virtual Platform of rare disease data and resources. Between 2004 - 2018 she worked on RD-Connect project, and contributed to the creation of flagship rare disease biobank data access tools. Internationally, Mary is actively contributing to operations of IRDiRC and planning of global RD roadmap. In addition, she participates in the International Consortium for Personalised Medicine (ICPerMed) Executive Committee, and is leading a workpackage in the IC2PerMed project on engaging China on PerMed research. Mary received her PhD from University College London.

Ma'n Zawati



Ma'n H. Zawati (LL.B., LL.M., Ph.D. (DCL)) is an Assistant Professor at McGill University's Faculty of Medicine and the Executive Director of the Centre of Genomics and Policy in the Department of Human Genetics. He is also an Associate Member of McGill's Biomedical Ethics Unit. His research concentrates on the legal, ethical and policy dimensions of health research and clinical care, with a special focus on biobanking, data sharing, professional liability, and the use of novel technologies (e.g. mhealth apps, WGS, WES) in both clinical and research settings. Dr. Zawati has published 13 book chapters and 45+ peer reviewed articles in leading publications.

