CVBF
Consorzio per Valutazioni Biologiche e Farmacologiche

CVBF ACTIVE INVOLVEMENT

IN THE EUROPEAN JOINT PROGRAMME RARE DISEASES

The **European Joint Programme on Rare Diseases** (**EJP RD**) is aiming to creating an **effective rare diseases research ecosystem** for progress, innovation, and benefit of everyone with a rare disease (RD).

To do so, the Programme supports RDs stakeholders by *funding research*, bringing together *data resources and tools*, providing dedicated *training courses*, as well as *translating high quality research into effective treatments*.

Since the beginning, CVBF has had an active role on several EJP RD work packages (WPs) focused on highly relevant issues around the RDs ecosystem.

WP1 Coordination and management

CVBF is involved in the **monitoring process** set within **WP1** aimed to evaluate the **quality of the EJP RD activities** and the **achievement of the expected project goals** during its whole duration. The EJP RD monitoring process is based on specific metrics such as *key results indicators* (KRIs) and *key performance indicators* (KPIs). ***CVBF*** develops annual Monitoring reports based on the information gathered by the EJP RD partners. The monitoring activities and reports support the assessment of the EJP RD advancement towards specific impacts set for the Programme.

WP2 Integrative research and innovation strategy

CVBF is involved in this **WP2**, where it worked on **Task 2.1** "**Prioritisation Scheme**" for EJP RD actions. Under this task, ***CVBF*** contributed to the creation of the Prioritization Guidelines, which are based on criteria and procedure specifications for decision-making processes on needs and actions to fulfill the EJP RD objectives.

WP4 Ethical, regulatory, legal and Intellectual Property Rights (IPR) framework of the EJP RD

In this WP, ***CVBF*** is part of the **Advisory Regulatory Ethics Board (AREB)** that oversees ethical issues and regulatory procedures linked to the EJP RD activities.

CVBF CVBF contributes to ensure compliance with all international and European relevant rules and ethical norms, including but not limited to fundamental and patients' rights, data management, data protection and confidentiality, related to the projects founded within EJP RD and EJP RD itself.

WP16 Online Academic education course

The EJP RD **Massive Open Online Courses (MOOC)** are designed as part of this WP16 to facilitate the access to trainings on RD research advances. Specifically, in the WP16 framework, ***CVBF*** is developing the ***MOOC-2 "Innovative Therapies and Personalised Medicine: New keys for the treatment of rare diseases"***, in collaboration with the *French Foundation for Rare Diseases* and *ERN Transplant Child*.

In particular, this **MOOC-2** course aims to provide an overview on the different innovative therapeutic approaches developed in the context of RDs as well as catches on the ethical, economic, and legal challenges related to their implementation.

MOOC-2 is organised as a journey, which covers different topics distributed in a 5 weeks programme and it is based on asynchronous content (i.e., the attendant can learn at his/her own pace). It has been developed in 2021 and in the first semester of 2022, to be ready for the launch at the end of 2022.

CVBF is responsible for the preparation of *"Personalised/precision medicine*" (week 4) and "*Marketing and legal challenges related to the implementation of innovative and personalized therapies*" (week 5). Several experts have been involved in the development of the MOOC contents, which delivered in different formats (i.e., text, cartoon, power point, interview, testimony and so on).

Indeed, weeks 4 and 5 provide an insight on the current innovative therapies landscape and the actors involved, including the regulatory, IP, and market implications. The participants will be able to learn more about why personalised medicines are so important, the current strategies of their development as well as the role of the artificial intelligence and predictive tools in the evaluation of the health risk and design of personalised medicines.

CVBF's partners that are currently working on the development of the MOOC's topics are: *Duchenne Data Foundation*, *Biomedical Research Foundation of the Academy of Athens*, *University of Bari 'Aldo Moro'*, *IRCCS Burlo Garofolo*, *Telethon*, *TEDDY Network*, *The Cyprus Institute of Neurology & Genetics (CING)*.

MOOC-2 is primarily targeted to students in *medicine*, *biology*, or *life sciences*, young researchers (PhDs and Post-Docs) in these disciplines, as well as healthcare professionals who want to learn more about innovative therapies and personalised medicine in RD. However, the language used is made understandable to everyone with a minimum of scientific education.

WP17 ERN RD training and support programme

To increase the global scientific and technological capacity in Modelling and Simulation (M&S), ***CVBF*** in collaboration with the *University of Bari 'Aldo Moro' (UNIBA)* and *University College of London (UCL)* as part of **WP17**, organised the workshop ***'Modelling & Simulation: Research Methodologies for Small Populations in Rare Diseases'*** that was **held in Bari**, **Italy on 4-5 July 2022**.

The workshop aimed at facilitating the discussion and the knowledge exchange on modelling and simulation methodologies as well as strategies as innovative and promising enough for facing complex multifactorial or RDs and conditions that require highly specialised treatments and resources. As such, the workshop addressed specific issues on RD for small populations.

More than 20 researchers coming from Institutions affiliated to one or more of the 24 *European Reference Networks (ERNs)* participated in the Workshop both in-person and remotely. The training methodology was based on lectures, seminars, and practical sessions, to provide concrete research skills.

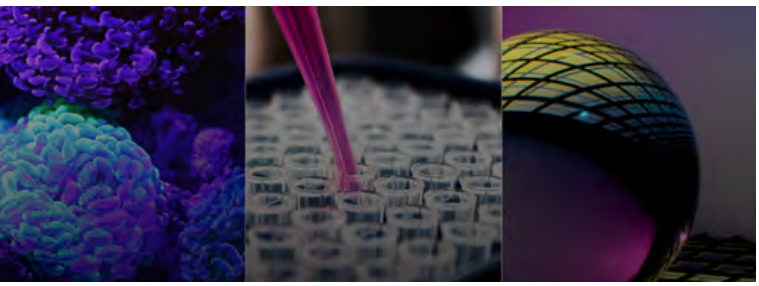
Specifically, **real-life case studies** were discussed during interactive and user-friendly seminars where the attendees had the opportunity to **apply knowledge using available databases and software**.

The Workshop Agenda foresaw three main topics:

- **Paediatric toxicology** on application of web platforms for in-silico assessment of toxicity and the utility of a machine learning approach for the prediction of developmental toxicity of chemicals covered by UNIBA
- **Regulatory part** on what is needed for translational preclinical studies covered by CVBF
- **Clinical part** on a) the utility of PK/PD modelling and b) the simulation in drug development for small populations covered by UCL

> ***WP19 Facilitating partnerships and accelerating translation for higher patient impact***

CVBF brings paediatric competences and expertise within the **WP19** thanks to its coordination role of the *European Paediatric Translational Research Infrastructure (EPTRI)*.
In particular, ***CVBF*** actions within this work package are aimed to support the RD community to translate high quality research more effectively into valuable impact interventions for the RD patient community. The **final goal** is to create a bridge between *basic research* and *medical innovation*, also known as the "bench to bed-side" approach.



To reach this goal, ***CVBF*** and WP19 Partners have developed the ***Innovation Management Toolbox (IMT)***, an online virtual library that provides self-help resources in RD translational medicine openly accessible to the whole RD research community.

The ***IMT*** empowers researchers to conduct rigorous translational research. It supports researchers to independently navigate the complexities of translation and give a clear overview of the communities that are available.

The ultimate impact of such activities is to reduce uncertainty and inefficiency in the academic translational research process, resulting in faster development and more patient-centric research outcomes.

The *toolbox* includes:

- technical guidance (such as target validation techniques, use of molecular imaging for drug development, patient confidentiality and data handling, animal welfare, biorepositories, intellectual property management and contract templates)
- a project management manual
- a regulatory guidance to support researchers in identifying key issues during the innovation translational research process

Moreover, ***CVBF*** provides support on aspects related to paediatric pre-clinical research and to the assessment process within project mentoring and technical support offered by WP19.

> *The CVBF's team in EJP RD is composed of young, professional, and highly motivated people.*

Donato Bonifazi,
Translational Innovator

'Many rare diseases develop during the infancy lead to death or permanent damage due to missing or malfunctioning elements, be them genes or cells. Treatments with exogenous replacement substances are very hard in these conditions as the dosage and schedule of administration needs to be continuously adjusted. Instead, the insertion of a correct sequence leads to self/regulation and most important to a single treatment in life freeing from the constrain and dependence of continuous monitoring.'

Giorgio Reggiardo,
Clinical Trial Statistician

'My contribution is to describe in short pills the journey from the study protocol design to the statistical analyses in the rare diseases domain, with the aim of applying the most innovative methodologies in clinical trials from adaptive design and big data analysis to innovative and precision therapies. As biostatistician, I propose statistical methods to establishing optimal treatment rules for personalised medicine.'

Bonka Georgieva,
Project Manager

'I am proud to represent CVBF in the EJP RD programme and to work with such committed EJP RD partners to make a difference for RD patients. Like a living puzzle, slowly but gloriously, every added piece leads us to discover what the future of the RDs research ecosystem is to be.'

Claudia Pansieri,
Health Data Scientist

'Rare diseases undergone extraordinary strides forward in terms of scientific research, technology, and capacity building. Thanks to the close collaboration with the leading experts in this field, in the EJP RD programme, we have access to the most innovative research in RDs as a unique common European health data space.'

Giovanni Migliaccio,
Regulatory Expert

'For me is an honour to provide advice and expertise as a Member of the EJP RD Advisory Regulatory Ethics Board, by analysing of the most relevant results the project obtained and supporting the development of innovative tools and methodologies for drug discovery and development in the field of rare diseases. We all work to overcome the fragmentation that still exists in the rare diseases as well as in the paediatric research, and to reduce delays in diagnosis and in access to available care.'