ERICA

European Rare Disease Research Coordination and Support Action

ERICA 2nd General Assembly 20-22 June 2022, Bologna, Italy

SUMMARY REPORT

The **European Rare Disease Research Coordination and Support Action- ERICA 2nd General Assembly** took place in Bologna from June 20-22, 2022, as a hybrid meeting with more than 100 people attending in person and online. Hosted by Istituto Ortopedico Rizzoli (IOR) and ERN BOND Coordinator Luca Sangiorgi.

The aim of the ERICA consortium, in which all 24 European Reference Networks (ERNs) take part, is to build on the strength of the individual ERNs and create a platform that integrates all ERNs research and innovation capacity. This symposium was a great opportunity to discuss the progress and future of the ERN related Research activities and to participate in the WP-Specific Expert Working Group sessions to exchange the success stories and to brainstorm with the best experts on the field.

All the presentations are available at ERICA website: https://erica-rd.eu/about/governance/general-assembly/ga2022/presentations/

And at ERICA SharePoint (including videos and photos): ERICA 2nd GA 20-22 June 2022 Bologna (hybrid)

ERICA has received funding from the European Union's Horizon 2020 research and innovation programme under grant agreement No 964908

Work Package's (WP) Summary remarks.

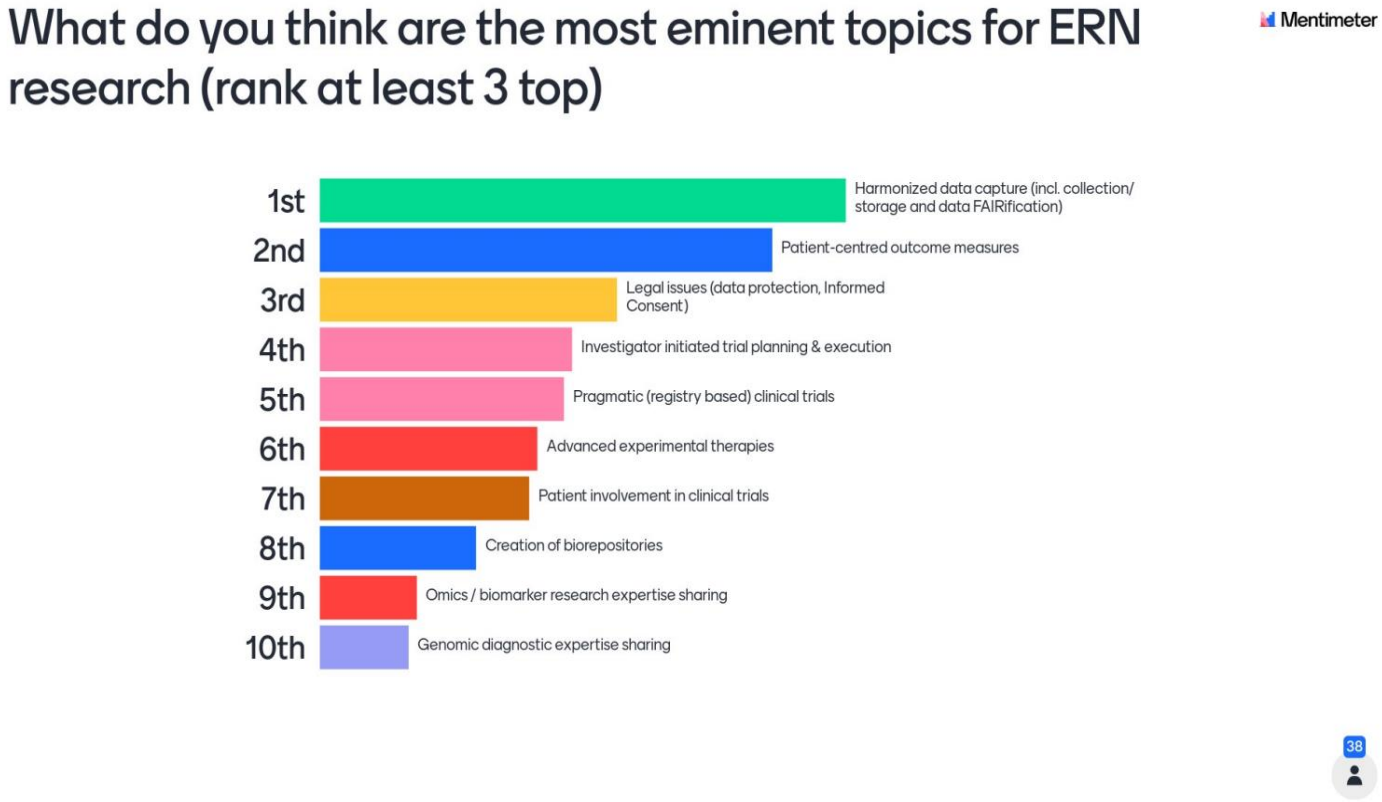
WP1, WP2, WP3, WP4, WP5, WP6

Consortium management and coordination (WP1)

General presentation of the project *Alberto Pereira* (ERICA coordinator) link

During the symposium we took opportunity to test the same questions that were asked few years ago within ERN Research WG, that formed a bases of the ERICA objectives.

The **'strengthening of the ERN Research Collaboration'** came out again as the top general priority of the ERICA and the interactive Mentimeter session also demonstrated that the most eminent topics for ERN Research are still dynamic and represented in the ERICA Work Packages.



The first attempt to define **the Common ERN research Roadmap** has been made by describing how the activates we envision in ERICA strengthen the activities of the EJP RD, and how we all together will form a very important part of the framework of the RD Partnership programme. ERICA and ERNs have a central role in Rare Disease Research ecosystem.

So far all the deliverables and milestones are on schedule and submitted on time.

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Data Collection, Integration and Sharing (WP2)

ERICA WP2 conducted a four hours interactive session on the **Rare Disease Partnership Strategic Research and Innovation Agenda (SRIA)**.

The first part of the discussion was dedicated to the **ERNs research priorities** that need to be included in the SRIA. Discussions were conducted using a Mentimeter survey. The audience was mainly constituted of ERN researchers and project managers, the opinion of patients associations and patients representatives is therefore not well depicted in the results presented below.

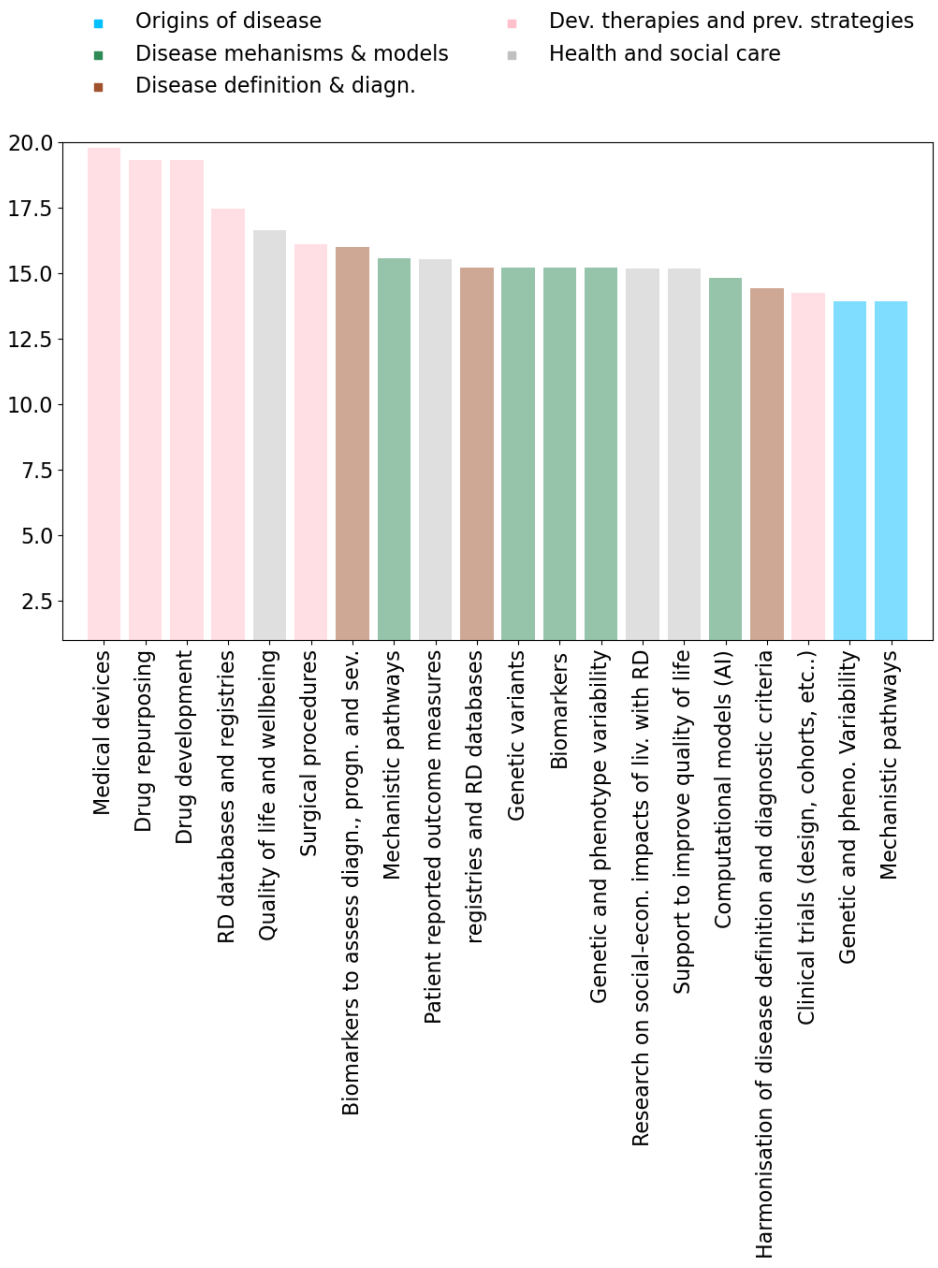

Figure 1: The 20 most important research priorities, as determined by the attendees of the ERICA GA 2022 in Bologna, Italy.

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The attendees were first asked to grade the importance of general research topics from 1 to 5, and, within these general topics, to grade the importance of specific research topics from 1 to 5. The 20 most important research priorities, classified by general research topics are depicted in Figure 1 above. It can be observed that the **most important general research priority is the development of therapies and preventive strategies.** To gain more confidence in the results, it is suggested that every ERNs and EURORDIS run the same type of survey within their own community.

The second part of the discussion was dedicated to the **Clinical Research Network (CRN)** that will be a central part of the RDP. The following main points were raised:

- The support services that will be provided with the CRN need to be tested with use cases before the deployment of the CRN to make sure that they are fit-for-purpose.
- It will be crucial to provide an AI support service, especially to provide support for the design of studies involving AI.
- Data exploitation hub is about the re-use of the data, but the way it will operate and its governance needs to be clarified.
- Clinical trial support will extend the existing services of EJPRD (methodology, design, clinical readiness) but it is feared that this is not going to be enough. It needs to be attractive for the industry to join, but not overlap with the work conducted by IHI. One aspect of the clinical trial support would be to focus on drug repurposing, which is of low interest for the industry.
- Diagnostic support services will focus on epigenetic, variants of unknow concern and phenotyping. It should make the link between the different existing models/experts across different institutions and work towards developing optimisation guidelines of diagnostic procedures.
- ePAGs would like to see a specific service dedicated to them that would provide translation services in different languages and support patient engagement in clinical activities.
- Overall, longer discussion with all the stakeholders will be needed once it becomes clearer what the CRN and its support services will be.

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Patient-Centred Research (WP3)

The **WP3 "Patient Centered Research in Rare Diseases**" aims to **define priority areas for future Patient Reported Outcome** Measures (PROMs) **development, support ERNs in the implementation of PROMs** and to **create a central repository of PROMs for ERNs.**

Objective and results achieved so far: The first version of the central repository of PROMs for ERNs has collected **672 PROMs:**

1. 317 were developed for Rare Diseases (RD)
2. 5 were generic Quality of Life instruments
3. 200 focused on Orphanet Disability Questionnaire (ODQ) functional consequences
4. 150 additional PROMs have been identified via the ERNs Survey

How ERNs can contribute to WP3:

1. Answering the ERN Survey ,

2. Annotating/Updating a RD by participating in a disability interview (Interview questionnaire available here and here), by contacting disability.orphanet@inserm.fr

3. Being a beta user for testing the PROMs repository and

4. Getting access to PROQOLID by contacting celine.desvignes-gleizes@mapi-trust.org

Next steps are:

1. Provide each ERN with a compiled report of available PROMS and GAP analysis and

2. Identifying pilots for PROMs implementation and/or validation

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Clinical Trial Support (WP4)

Deliverables we are currently working on:

D4.2 Report on prepared factsheets and YouTube movie (M36)
D4.3 Software wizard for CT support developed (M36)
D4.4 Report on frequency of use of the website and usefulness of the software wizard (M36)
D4.5 Report on the outcome of clinical trial workshops (M48)
D4.6 Report on achievements of ERN-EMA dialogue (M48)

Webinars, Factsheets and YouTube Movies

In order to produce effective factsheets and YouTube videos we have decided to set up a procedure according to whom a preparatory webinar involving different stakeholders will be performed and according to the discussion the factsheets and the video will be developed

These are the webinars that have been defined:

- **Essential requirements before thinking about a clinical trial**

Viviana Giannuzzi, Benzi Foundation

- **Feasibility for clinical trial – what Pharma Companies expect and what they require as indispensable**

Diego Ardigò, Chiesi

- **Definition of orphan drug by the EMA**

Armando Magrelli, ISS

- **REMEDI4ALL and expected impact for rare diseases clinical trial**

Anton Ussi, EATRIS

- **The impact of PROMs and PREMs in the preparatory phase of a clinical trial**

Ana Rath, INSERM - Manu Mar, VHIR

- **Framework for Patient Engagement in clinical trials**

Maria Cavalier, Virginie Hivert

During the meeting further inputs through Mentimeter have been collected from the participants

We will also continue the **Collaboration with WP2 on biobanking** specifically on these points:

- Preparation of the biobanks survey for the ERNs HCPs
- Discussion of the results and definition of a strategy

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Translation and Innovation (WP5)

WP5 provided an overview of the performed activities in the past year, such as the establishment of the Innovation Expert Working Group (I-EWG). The I-EWG is composed of ERNs clinicians and researchers, external experts, ERICA Innovation advisory members as well as patient representatives. Their main task is to create a roadmap and strategic plan for accelerating innovation and translation of research in ERNs. This will be delivered through mapping of educational and research needs amongst ERNs (survey recently held) as well as by organizing educational webinars (first one held in Nov 2021) and educational and innovation workshops. At the kick-off meeting held in Feb 2022, the I-EWG discussed the outcome of the survey to help cluster and prioritize the main bottlenecks, showcase success stories, and identify potential solutions to support ERNs in bringing innovations closer to the patient. This led to the first Innovation Workshop, organized during the ERICA GA in Bologna within WP5 dedicated session on Wednesday 22nd June from 11:00 – 13:00. Invited experts from different ERNs presented the most inspiring success stories on the topics of drug repurposing, preclinical research, and trial design in rare diseases. WP5 is grateful to Gilles Vassal, MD, PhD (ITCC, ERN PaedCan), Bas Vastert, MD, PhD (UMC Utrecht, Wilhelmina Children's Hospital ERN RITA), Jørgen Mogens Thorup, MD, PhD (Uni Copenhagen, Department of Clinical Medicine, ERN eUROGEN), Valentina De Giorgis, MD, PhD (Struttura Complessa di Neuropsichiatria Infantile Centro di Epilettologia dell'Infanzia e dell'Adolescenza , ERN EpiCare), and Loes van der Zanden, PhD(Radboud University medical center, Department for Health Evidence, ERN eUROGEN) for accepting the invitation and sharing their knowledge and experience with the RD community.

This was an important step forward in connecting ERNs and sharing good practices for the benefit of future translational research projects. The I-EWG will continue to work on supporting ERNs in their innovation and translation potential. The next meeting is foreseen to take place in Vienna this Autumn.

WP5 also had the opportunity to showcase the EJPRD Innovation Management Toolbox (IMT) and ERICA Catalogue of services (CoS) which will be integrated in the IMT. The IMT is a reference library of resources, tools and services available to the rare diseases community. It is free to use and has been designed to provide investigators with self-help resources specific to their needs. The IMT is officially launched on June 30th 2022.

The integration of the CoS in the IMT will stimulate cross fertilization between both EJPRD and ERICA project and provide a single place for researchers to find and access all necessary rare disease tools, services and resources. Furthermore, the IMT has some great functionalities such as a search and browse function and smart filtering using tags.

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Integration, Outreach & Dissemination (WP6)

ERICA Website https://erica-rd.eu/ is operational and is becoming increasingly interactive. We will soon harbor a specific web platform that depicts the ERN Research activities such as Clinical Practice Guidelines (CPGs) and Clinical Trials (CTs), that are aligned with Orphanet database.

Main aim is to structure the Orphanet Database (DB) the way that ERNs can take best use of it and extract data for the ERN research purposes. The DB created for ERNs is not in competition or duplication with other databases, but it can be discussed further if ERNs would like to deposit and control their data in Orphanet and let Orphanet make the extractions for the websites of ERNs.

In return ERNs can show their activities through Orphanet (more than 2milj users per month) and combine all the ERN activities within ERICA. Within ERICA we will create an ecosystem that structures the data and not only displace the data.

Added value of the ERICA is to show the ERN capacity of research (via Orphanet Rare Disease DB) and also to help ERNs to list their research data (e.g. for Monitoring).

During the first exercise 194 Clinical Practice Guidelines connected to specific ERNs were detected, that will get additional 'ERN Tag' according to Continuous Monitoring (CMP) definition.

To validate Clinical Trials (CTs) that can be associated with specific ERNs, it was decided to broader the CMP definition and include also the trials that are only conduced in one member state, as long as conducting centre is part of ERN.

The centres in HCPs are already in Orphanet and we are crossing the data between CTs and ERNs, trying to find the CTs that are happening in the centre that belongs to a ERN. That is the part that might not match very well.

- Proof of Concept is tested with the Metab-ERN and Endo-ERN to see if the datafile with candidate trials is easy to understand, easy to work on it etc. After feedback from Endo and Metab, Orphanet will analyse the results that will allow to tag in the Orphanet DB the ERN CTs according to definitions discussed (*NB! The CTs that are done by ERNs (members/centres). ERNs do not conduct(fund) CTs!*). Once the exercise is done, we will scale up to all ERNs and send them the candidate list of CTs for you to check from ERN perspective weather you miss any data.
- MetabERN proposed to check that all centres running these CTs are certified to run the relevant phases (I , II, III, IV), and is currently exploring this option for their ERN data to see if it is feasible for the this exercise across all ERNs, or if it should be part of the ongoing improvement efforts of the DB once published.

This task shows also if we are depicting all the ERNs CTs, if we are missing a lot (and why) , if we are duplicating or if we could do something more.

Main conclusions of the session-

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Need to better define the objectives of the task and what we want to do with the repository:

- Need to clearly define the criteria.
- To clearly define what aims we are pursuing with this exercise.
- Need to emphasize that the data is provided by Orphanet to the ERNs (no extra gathering of data is required).

We pursue the aim: To have platform for patients and for all that are involved in RD research, where they can go and identify the trial as RD research or trial (however u define it) for the sake of the patient, but we need to be careful how we phrase our messages and what efforts we undertake for what reason. Important to consider what it means to raise hopes for the patients, when you do not have funding to transfer them cross-border.

Suggestion- to add into proof of concept also the 'Impact Assessment' (for the funding, patients, etc.).

Agreed- Before going public, we need to take time for reflection.

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