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REPORT FROM MEETINGS OF THE COORDINATION ACTIVITY

Executive Summary

One of the tasks of ADOPT WP7 dealing with Rare Diseases (RD) is to coordinate the efforts and integrate existing biobanks and resources within the RD field towards sustainable biobanking. Important for achieving this task is the alignment of activities within RD projects and initiatives to ensure the sustainable base for the RD biobanks within BBMRI-ERIC. This objective has been met by participation to various meetings with the RD operators and representatives (e.g. RD-CONNECT, ERN BOND, ERA-Net EURODIS etc.), and discussing, as well as executing, the future collaboration with the National Nodes working towards to the establishment of the Common Service for Rare Diseases. Networking has been realized particularly with BBMRI-ERIC and RD-CONNECT in order to provide sustainable cataloguing solutions for the RD-CONNECT within BBMRI-ERIC.



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1. Background

RD research projects have developed many biobanks and registries in Europe as part of their efforts to allow access to samples and data for research and clinical trials. Several projects have also contributed to the RD area in providing shared tools and unified platforms for the benefit of the RD community. However, many initiatives suffer from unsustainable structures for governance and funding schemes. Part of the work on ADOPT WP7 is to coordinate and integrate the activities and efforts of different projects involving RD biobanks. For this purpose, BBMRI-ERIC, LUMC/UMCG and IOR together follow the developments within the RD community, participate in meetings and coordinate working group teleconferences for the advancement of sustainable base for biobanking activities within the RD community.

2. Approaches (Methods)

This report is an update to the reports provided at M12 and M24 including information on several meetings, teleconferences and a webinar relevant to the coordination activities of WP7 in the RD field.

3. Results

3.1 Coordination activities (M1;Oct 2015- M12; Sept 2016)

Several meetings and teleconferences have been attended by BBMRI-ERIC, LUMC/UMCG and IOR representatives under the coordination duties (Table 1). The 1st Coordination meeting was held in November 2015 (Table 1). The focus of the meeting was to provide an update of any news relevant to the RD Community and to draft a plan for the upcoming H2020 calls (participants present from BBMRI-ERIC, LUMC/UMCG and IOR). An important collaborative meeting with RD-CONNECT representatives was held in March 2016 during the RD-CONNECT Annual meeting within a separate session on ‘sustainability of key RD-CONNECT assets’. This session determined the future collaboration steps and the sustainability plan aimed at continuing the cataloguing efforts of RD-CONNECT within BBMRI-ERIC (participants from BBMRI-ERIC and IOR). In May 2016, a coordination teleconference was organised with several research infrastructures (EATRIS, ECRIN, BBMRI-ERIC, ELIXIR) to discuss the joint programme for RDs and to discuss the national strategies towards the joint effort in RD field. Table 1 addresses the other meetings in regards to the coordination efforts in RD field realised within the 1st year of ADOPT BBMRI-ERIC.

Table 1.

Meeting nro	Name of the meeting	Participants	Locations and dates	Meeting
1	<i>Kick off Meeting ADOPT BBMRI-ERIC</i>	L. Sangiorgi, GJv Ommen, O. Törnwall, JE. Litton	<i>Vienna, 20-21/10/2015</i>	<i>Kick off Meeting</i>
2	RD CONNECT ANNUAL MEETING	L. Sangiorgi, M. Mordenti, GJv Ommen, O. Törnwall,	Barcelona, 8-10/03/2016	Annual meeting



3	EUROBIOBANK Assembly meeting	L. Sangiorgi,	Barcelona, 9/03/2016	Assembly meeting
4	ADOPT BBMRI ERIC MEETING	L. Sangiorgi GJv Ommen, O. Törnwall, JE. Litton	La Valletta Malta, 16-18/03/2016	Project meeting
5	ADOPT BBMRI ERIC AoM#6	L. Sangiorgi	Vienna, 27- 28/04/2016	meeting
6	ECRD 2016 : The European Conference on Rare Diseases & Orphan Products	L. Sangiorgi	Edinburgh 26- 28/05/2016	Meeting-Poster
7	RD-ACTION Workshop: 'Exchanging data for virtual care within the ERN Framework	L. Sangiorgi	Brussels 28- 29/09/2016	Workshop
8	Europe Biobank Week, Patient and ELSI sessions.	L. Sangiorgi	Vienna, 13- 16/09/2016	Meeting



3.2 Coordination activities (M13; Oct 2016- M24; Sept 2017)

During this period, BBMRI-ERIC has participated at several meetings to make the BBMRI-ERIC IT services interoperable and aligned with the services of the RD Communities, namely represented by RD CONNECT (Table 2). BBMRI-ERIC also works towards the sustainability of RD CONNECT (e.g. integrating the ID cards within the BBMRI-ERIC Directory) and facilitating access to RD biobanks by specific tools (e.g. BBMRI-ERIC Negotiator).

IOR has also been leading the coordination efforts on their behalf by partaking and presenting ADOPT project activities in several meetings relevant to the RD field (Table 2). During this period, IOR in collaboration with ERN BOND (ERN on bone disorders), mainly focused on setting up a collaboration for future joint activities with Osteoporosis Imperfecta Federation Europe (OIFE) and Brittle Bone Diseases Consortium (BBDC). As reported at M12, ERN BOND has settled the collaboration with OIFE for the conduction of a survey on the state of the art of diagnosis and treatment of OI in Europe.

The Objective of the Survey

The aim of this questionnaire was to provide a picture on the situation in the 10 Member States represented within ERN-BOND (Belgium, Czech republic, Estonia, France, Germany, Italy, The Netherlands, Portugal, Sweden, United Kingdom) in regard to diagnosis and to identify the main challenges and potential solutions that could help reduce diagnosis delays. This activity served to identify existing care gaps within and among ERN BOND full members and to collect patient information on their experience leading to the disease diagnosis. The results of the survey have been reported into a White Paper (<http://brittlebone.org/oi-white-paper-ern-bond/>) that shed light on the unmet needs of patients in this specific area and will serve as a basis for a dialogue with policy-makers on the challenges related to diagnosis of rare bone diseases..

This questionnaire, focusing on Osteogenesis Imperfecta (OI), one of the disease areas covered by ERN BOND, was used as prototype taking into consideration that OI represents a heterogeneous group of generic disorders that affect the bones.

OIFE facilitated the capillary dissemination of the questionnaire among patients, utilizing the OIFE Network in Europe.

Lastly, a member of OIFE is an ePAG (patient representative) in ERN BOND and is member of the BOND Steering Committee, the most important organ in the governance of the Network.

Table 2.

Meeting nro	Name of the meeting	Participants	Locations and dates	Meeting
9	<i>RD CONNECT Hackathon</i>	P Holub, D.J. van Enckevort	<i>Milan 22/11/2016</i>	<i>Sustainability and synergies</i>
10	<i>RD CONNECT & BBMRI-ERIC sustainability meeting</i>	P Holub, D.J. van Enckevort	<i>Milan 20/02/2017</i>	<i>Sustainability and synergies</i>
11	<i>BBMRI & European Commission's Joint Research Centre (JRC) meeting</i>	P Holub	<i>Ispra 13-14/06/2017</i>	<i>Collaboration with BBMRI and JRC</i>



12	Kick off meeting ERN BOND	L. Sangiorgi	Rome, 22-24/05/2017	Kick off Meeting
13	Meeting with OIFE	L. Sangiorgi, I Westerheim, R Tvedt	Oslo, 26/08/2017	Future collaboration with OIFE and ERN BOND
14	OIOslo2017, 13th International Conference on Osteogenesis Imperfecta	L Sangiorgi, I Westerheim, R Twedt, L W Valerie, C Daire, K Javaid	Oslo 27-30/08/2017	Collaboration meeting with OIFE and ERN BOND
15	MonitoRaRe - Report 2017	L Sangiorgi	Italian Parliament Rome 27/07/2017	An update on the Italian national and European situation on RDs.
16	Brittle Bone diseases Consortium (BBDC)	L. Sangiorgi, C V R Sutton	Oslo 27/07/2017	Towards structured collaboration of ERN BOND and BBDC
17	13th meeting of the International Skeletal Dysplasia Society	L. Sangiorgi, C V R Sutton	Bruges 20-23/09/2017	Structured collaboration with BBDC, follow up meeting from BBDC (nro 13)
18	Meeting with CHAFEA	L. Sangiorgi, M de la Calle, P D'acapito	Luxemburg 06/09/2017	Future activitie discussions with ERN BOND and CHAFEA Project
19	Meeting with Lauro Panella and Antonio Tajani	L. Sangiorgi	Rome 07/09/2017	Presentation of the OIFE Survey White Book
20	EURORDIS	L. Sangiorgi	Brussels 07/09/2017	Preparation for presenting ERN BOND to during an upcoming Parliament audition in 2018
21	13th International Skeleltal Dysplacia Society (ISDS) Meeting	L. Sangiorgi	Bruges 20-23/09/2017	Presenting ERN BOND to ISDS
22	Second RD-Action Workshop "Using Standards and Embedding Good Practices to Enable Interoperable Data-Sharing in ERNs"	D.J. van Enckevort	Brussels, 27/04/2017	Recommended Practices for Data Standardisation in the Context of the operation of European Reference Networks
23	ADOPT mid term review	L. Sangiorgi	Brussels, 27-28/06/2017	Project meeting
24	BBMRI-ERIC assembly of members	L. Sangiorgi	Losanna, 19/05/2017	meeting



IOR has led and organised 4 teleconferences (15/11/2017, 06/02/2017, 10/05/2017, 05/07/2017) for the BBMRI Rare Diseases Working Group (WG) in order to sustain the continuous work on activities related to rare diseases. The WG teleconferences have involved representatives from BBMRI-ERIC, BBMRI.fi, BBMRI.pl, BBMRI.mt, BBMRI.be, BBMRI.ee, BBMRI.tr, BBMRI.nl, BBMRI.it, BBMRI.at and representatives from RD-CONNECT project. The WG efforts aim to avoid any duplication of work on overlapping activities among other linked projects ERNs, ADOPT WGs, RD-CONNECT and ELIXIR. The WG also enabled the involvement of the BBMRI National Nodes within the RD activities.

The main topics and achievements are as follows:

- Improvements and further development of the Helpdesk model
- Pilot Study on OI: collaboration with the National Nodes of BBMRI, the RD-CONNECT project and ELIXIR infrastructure towards:
 - o FAIR (findable, accessible, interoperable, reusable) data within the biobanks and RD registries
 - o ERN BOND involvement
 - o Mapping of data and samples for OI (what is available and where) based on the data model of IOR has been initiated. Support for this work will be provided by ELIXIR and BBMRI-ERIC Common Service IT.
 - o Current involvement of 250 patients for OI with further cases in the pipeline
- Steps taken towards assessing the sustainability of RD biobanks, how to move forward e.g. collaboration with Pharma and ERNs -> small working group to be established for this.
- Upgrading the concept of RD Common Service by considering the existence of ERNs in EU – future users of BBMRI-ERIC and National Node services.
- Focus to invest in the quality of the sample related data
- The outputs of the Quality groups of BBMRI-ERIC and EuroBioBank will followed in regards to the sample quality
- The BBMRI-ERIC Directory of RD Biobanks, developed under BBMRI-ERIC by UMCG/LUMC will be tested also in the Telethon Network of Genetic Biobanks (TNGB)

RD Helpdesk Webinar

BBMRI-ERIC organised a Webinar on RD-Helpdesk on 16/06/2017 providing Hands-on tutorial showing how to use Request Tracking (RT) system¹, which is the basis for the Rare Diseases Helpdesk. The tutorial focused on how to submit a request (thus how to create a "ticket"), how to manage the ticket and also how to manage queues of tickets within the whole RT. The webinar was well received by the participants.

Patient involvement

BBMRI-ERIC has established a patient chapter as part of its Stakeholder Forum involving various patient advocacy groups (Genetic Alliance UK, Alzheimer Europe and Dutch VSOP) including a RD representation (EURORDIS). The aim of the Stakeholder Forum is to know and address the concerns of its stakeholders and to provide a platform for discussions for the various groups.

¹ <https://bestpractical.com/request-tracker>



3.3 Coordination activities (M25; Oct 2017- M36; Sept 2018)

Several meetings and teleconferences have been attended also during last year of project. (Table 3). BBMRI-ERIC works towards the RD CONNECT and regarding the RD CONNECT tool sustainability beyond the end of the project, a joint meeting with all entities involved (i.e., BBMRI-ERIC, RD CONNECT, EuroBioBank) has been held in September 2018 at IOR premises. Activities for the integration of RD CONNECT tools into BBMRI-ERIC platform are still ongoing.

Furthermore in order sustain the continuous work on the activities related to rare diseases, IOR has continued to lead and organize teleconferences (27/09/2017, 01/02/2018, 23/04/2018) for the BBMRI Rare Diseases Working Group (RDWG). The RDWG teleconferences have involved regular BBMRI-ERIC and RD-CONNECT representatives. The WG also enables the involvement of the BBMRI National Nodes within the RD activities: Representatives/contact person for National Nodes Members (Austria, Belgium, Estonia, Finland, Germany, Italy, Malta, The Netherlands, Poland, United Kingdom, Czech Republic, France, Greece, Norway, Sweden, Switzerland, Latvia) and observers (Switzerland, Turkey, Cyprus). Moreover Representative/contact person of Spain Biobanks and of Telethon Network of Genetic Biobanks have been involved.

The main topics and achievements are as follows:

- OI Pilot study: to make data of biobanks and registries Findable, Accessible, Interoperable and Reusable (FAIR) – a system developed at Leiden University, LUMC – a pilot study to demonstrate that it is possible to integrate data and make it interoperable:
 - selected ontologies: Orphanet (diseases), HPO (signs and symptoms) and HGNC (gene)
 - OI elements file (.xls) individuated from the data collection formats of OI registry, shared also with the RD WG
 - few driving questions for OI Pilot study, as example
 - Updating of the work progress by LUMC

- Helpdesk: preparation and discussion of the Frequently Asked Questions (FAQs) and Wikis, related to RD Biobanks and Registries

The Survey

The White Paper has been presented on the Rare Disease Day 2018 at the European Parliament. In the future, this questionnaire could be replicated in other disease areas.

Table 3

Meeting nro	Name of the meeting	Participants	Locations and dates
25	<i>ERN coordinators meeting</i>	Coordinator	Bruxelles, 10-12/10/2017
26	<i>FOP (Fibrodysplasia Ossificans Progressiva) Drug Development Forum</i>	BOND member	Alghero, 13-14/10/17
27	<i>ERN Info Day, Ministry of Health</i>	Coordinator	Rome, 16/10/17
28	<i>WORKSHOP ON HOSPITAL MANAGERS AND EUROPEAN REFERENCE NETWORKS IMPLEMENTATION</i>	L: Sangiorgi	ROTTERDAM 16-17 /11/17



29	<i>GIORNATA INTERNAZIONALE BBMRI.IT</i>	L. Sangiorgi, M. Locatelli	MILANO 15/11/2017
30	<i>BBMRI ERIC ASSEMBLY OF MEMBERS 10° SESSION</i>	L: Sangiorgi	vienna 8/11/2017
31	<i>Residential course "Congenital malformations: from prenatal diagnosis to postnatal therapy"</i>	L: Sangiorgi	ROMA 30/10/2017
32	<i>workshop on General Data Protection Regulation (GDPR)</i>	BOND member	Bruxelles, 20/10/17
33	<i>Hyperkalcemia and hypokalcemia in neonates, 19th Days of Pediatric Endocrinology</i>	BOND member	Pisek, 10/11/17
34	<i>Conference German Society of pediatric endocrinologists</i>	BOND member	Freiburg, 17/11/17
35	<i>Italian Society of Human Genetics Conference, XX Ed.</i>	project manager	Naples, 15-18/11/2017
36	<i>Workshop Role of Hospital Managers in Shaping the Future of the European Reference Networks.</i>	Coordinator, BOND member, HCP representatives	Rotterdam, 17/11/17
37	<i>Conference on Undiagnosed Rare Diseases, National Center for Rare Diseases</i>	project manager	Rome, 20/11/2017
38	<i>OSR meeting</i>	Coordinator	Milan, 01/12/17
39	<i>A girl with fracture of clavicle and lower limb, 18th Days of Pediatric Endocrinology</i>	BOND member	Pardubice, 01/12/17
40	<i>workshop on clinical practice guidelines, RD-ACTION.</i>	BOND member	Rome, 07/12/17
41	<i>ERN BOND Info Day</i>	Coordinator, project manager	Bologna, 18/12/17
42	<i>Workshop on the Preparation of Final Report</i>	project manager, financial manager	Roma, 22/01/18
43	<i>ERN Symposium – LUMC</i>	BOND member	Leiden, 24/01/18
44	<i>1st Meeting on Rare Bone Diseases</i>	BOND member	Coimbra, 16-17/02/18
45	<i>ERN BOND – First Technical meeting</i>	All BOND Members	Bruxelles, 26-28/02/2018
46	<i>ERN-BOND White Paper on Diagnosis, European Parliament</i>	All BOND Members	Bruxelles, 28/02/2018
47	<i>RE(ACT) CONGRESS 2018:</i>	Coordinator	Bologna, 07/03/18
48	<i>EPTRI Kick off Meeting</i>	L: Sangiorgi	ROMA 15-16 01 2018
49	<i>Meeting European Society on rare bone diseases</i>	L: Sangiorgi	16-17 1 2018 BRUXELLES
50	<i>EPJ RD MEETING</i>	L: Sangiorgi	PARIGI 5-6 2 2018
51	<i>RD-ACTION, EMA AND DG SANTE WORKSHOP: HOW ERNs CAN PROVIDE ADDED-VALUE IN THE AREA OF CLINICAL RESEARCH</i>	L: Sangiorgi	LONDRA 28-30 5 2018



52	<i>SYMPOSIUM IN CLINICAL GENETICS & GENOMICS</i>	L: Sangiorgi	ATENE 1-2 GIUGNO 2018
53	<i>X TRUST-4RD Methodology for use of RWE for Rare Diseases Stakeholder Roundtable</i>	L: Sangiorgi	21-6-2018 BRUXELLES
54	<i>RD ACTION FINAL EVENT</i>	L: Sangiorgi	28-6-2018 PARIGI
55	<i>EURO BIO BANK WHORKSHOP</i>	L. Sangiorgi, M. Locatelli	ROMA 18 10 2018
56	<i>9th WORLD ORPHAN DRUG CONGRESS</i>	L: Sangiorgi	BARCELONA 6-8 11 2018
57	<i>BBMRI AoM #12</i>	L: Sangiorgi	BREUKELEN 12-13 11 2018

3.4 Networking and Synergies

ERN on Rare Bone Diseases (ERN BOND)

The ERN proposal on Rare Bone Diseases composed by 38 Healthcare providers from 10 EU countries was approved in December 2016 by the European Reference Network Board of Member States. BOND combines skeletal dysplasias and metabolic bone diseases and the application has been coordinated by the ISDS, ESPE, ICCBH, ESE and ECTS. The Network Coordinator is Dr Luca Sangiorgi (IOR).

European Reference Networks (ERNs) create a clear governance structure for knowledge sharing and care coordination across the EU. They are networks of centres of expertise, healthcare providers and laboratories that are organised across borders. A centre of expertise could be a clinical team, a medical centre or a hospital and must be formally accredited by its Member State.

4. Discussion and conclusions

Through coordination of activities within the RD field, the WP7 is able to set out the platform for RD Common Service. Collaboration with various European projects and initiatives also enable to plan and execute the OI pilot study with maximum capacity and expertise. With coordinated actions, the tools developed in WP7 (e.g. Helpdesk, ID Cards for the RD biobanks) can be tested, validated and improved. Furthermore, transnational efforts executed in a coordinated manner are essential to optimally use the existing resources and avoid duplication of work.

5. Next Steps

Despite ADOPT project has come to the end, BBMRI Rare Diseases Working Group will continue to strength the established collaboration with other RD realities, i.e. BBMRI National Nodes, and ERN BOND. Several efforts will be put in place to continue towards harmonizing initiatives within the RD field, achieving sustainability and providing services for the RD community. ERNs is a pivotal collaborator and a high impact player in the RD field particularly as primary user of the services of BBMRI-ERIC and/or National Nodes especially the requests of samples and data access.



Appendix I: ERN BOND Newsletter N.1



ERN BOND Newsletter N.2

- ❖ **21st-22nd November 2018** – 4th Conference on European Reference Networks was held in Brussels. All presentations and streaming videos are available at the following link: https://ec.europa.eu/health/ern/events/ev_20181121_en
 - To see the promotional video animation on ERNs: https://ec.europa.eu/health/ern/videos_en
 - For those of whom participated to the Conference, you can give your feedback on the conference via this short survey: <https://ec.europa.eu/eusurvey/runner/ERNConferenceSatisfactionSurvey>. It will take only a few minutes and will be helpful for us to improve future events. The conference satisfaction survey will be available until 03/12.
 - A flash report on the Conference was published in the Commission webpage https://ec.europa.eu/health/sites/health/files/ern/docs/ev_20181121_sr_en.pdf
- ❖ **13th and 14th November** - Luca visited the Leiden University Medical Center to see the training and education platform in use at Leiden University and to new technology solutions that can be used within ERN BOND WG8 training plan.
- ❖ **c4c application** – BOND WG4 Clinical Trial is coordinating the application to c4c from ECRIN for funding from to undertake a study of repurposing losartan in osteogenesis imperfecta. <https://www.ecrin.org/news/c4c-call-proposals>
- ❖ **2019 Call for proposals on Registries** – European Commission confirmed a new call for registries will be come out in 2019 dedicated to ERNs. ERN BOND Coordinator will keep you updated.
- ❖ **15th December** – ERN BOND public website will be launched: <http://ernbond.eu/>
- ❖ **13th – 14 February** – the ERN BOND Annual Technical Meeting will be held, at Regione Emilia-Romagna Rue Montoyer 21 - 1000 Bruxelles. All ERN BOND Members are invited to participate.

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