ADOPT BBMRI-ERIC
GRANT AGREEMENT NO. 676550

DELIVERABLE REPORT

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FINAL REPORT

Executive Summary

The number of samples and associated data in the Rare Diseases field is rather limited, causing challenges in clinical trials and development of new treatments. The possibility to collect and share where possible was already agreed upon in several European Rare Disease projects, which all were funded during a limited time period. Adding the biobank activities of the Rare Disease community to BBMRI-ERIC could increase the awareness, visibility and secure sustainability.

Within BBMRI-ERIC, dedicated Common Services around Quality, IT and ELSI were created and at the start of the ADOPT project seemed a good model to use for Rare Diseases as well, but new insights show that a setup in a Common Service makes the straightforward need for support too complex and too much of an administrative burden. Therefore it has been decided to incorporate the RD activities in the overall program of BBMRI-ERIC and secure a custodian construction for already developed solutions and deliverables from other RD projects.

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Document log

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1. Background

BBMRI-ERIC was instrumental in defining what a modern, high-quality European biobank looks like, where supporting services were developed in the recent years to help achieve this, supported by ADOPT. These services are now reaching a mature status and can accelerate the pathway from laboratory discoveries to diagnostics and treatments. This requires integration in the real-life practice of academic and industry researchers, to ensure that these services become a real building block in the road towards new treatments. In addition, it is then crucial to operate towards a service-oriented total workflow model, serving the needs of all our stakeholders.

> *In the field of rare diseases (RDs) the number of available biospecimens is, in general, very limited. As a direct consequence of disease rarity, clinical trials are difficult to perform and so a limited number of treatments have been developed, whereas disease prognosis and natural history are poorly known, and patients with RDs do not receive the care and medical attention available to people with common diseases. Sharing material and data on RDs is essential for identifying disease-causing genes, studying pathological mechanisms, and developing treatments. (European Journal of Human Genetics, 2014, M. Mora et all.)*

The uniqueness of Rare Diseases and the crucial role that biobanks could play in the development of new treatments have led BBMRI-ERIC to participate in several European projects focussing on Rare Diseases. Many RD research projects (e.g., RD-CONNECT, other FP7 projects and the Joint Research Centre[1]) and networks (e.g., TREAT-NMD[2]) have developed shared tools and unified platforms on either biobanks or registries, or both. They have collected biomaterials and data to allow accessibility for research and clinical trials, but the conjunction of biobanks and rare diseases-oriented registries to

[1] https://ec.europa.eu/jrc/
[2] http://www.treat-nmd.eu/

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pool together samples and their clinical information remain a critical issue, including the funding to continue.

With this in mind, BBMRI-ERIC could act as a sustainable custodian for deliverables related to Rare Disease that were developed and launched in numerous projects in the past years. The role of custodian was discussed during AoM meeting #12 in Vienna, and a clear wish to go this route, starting with Rare Diseases, was agreed upon. Only by combining efforts and decreasing fragmentation can the Rare Disease Community have an impact. Through a Research Infrastructure sustainability can be guaranteed, truly serving the patients in the end.

As part of the efforts to support the Rare Disease biobanks the goal was to establish a dedicated Common Service giving information on quality standards, data collection, ethical and legal requirements.

2. Approach (methods)

BBMRI-ERIC already developed a Common Service ELSI and Common Service IT to serve the needs of the community with specific questions and/or issues. The idea was to have the same approach for Rare Diseases and secure in this way the opportunity to serve the community, while also guaranteeing the fact that National Nodes are involved in the execution and management of the specific services. Once a new structure is agreed upon, the various deliverables from WP7 will be integrated into the overall common services for Rare Diseases.

Since the Quality services are organized in a different, more top-down, approach this model will also be explored as a potential way forward.

3. Results

In the first period of ADOPT the individual needed services were developed for the Rare Disease biobanks, like the Helpdesk and the addition of the RD samples in the directory. Once finalized they could be hosted and delivered through a new common service, but with the change in strategy after summer 2017 to a more service-oriented approach, also the setup of the Common Services was reviewed.

The review of CS ELSI started with an internal assessment and a series of analyses to identify strengths, weaknesses, opportunities and threats (SWOT). Among the issues noted, it is stated that the ELSI experts recruited in each Member country are not necessarily linked to the National Node and/or aware of its activities and identify themselves with BBMRI-ERIC as an organization (e.g. via subcontracts, consultancies or project related). It is also stated that there is a lack of understanding that a Common Service ELSI is to be based on the user needs of the biobanking and biomolecular resources community. Many of the other weaknesses identified in the SWOT analysis reflect the need to better set and manage priorities, then budget and manage resources according to these priorities. One important threat that was identified was: "*Focusing on issues that are not pertinent to end users but to the academic forthcoming or project deliverables.*" The following are other weaknesses identified in the SWOT analysis that reflect the need to better set priorities and manage resources:

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- Lacking time and resources to engage in basic discussions in defining where harmonization is desired and what the process to reach it is
- Fragmentation of CS ELSI due to high number of contributors with few FTE and unclear mandates
- Poor ability to utilize additional project funding effectively (needs amendments of projects to bring in new institutions - either as linked third parties, or even full beneficiaries, because not everybody can have the linked third party status)
- Lack of time, resources, and processes to exchange and collaborate efficiently across BBMRI committees and bodies for developing BBMRI-ERIC tools, services and policies in a constructive and timely manner.

Three external reviewers used the above analysis as a starting point and reported in an internal report to the Assembly of Members in November 2018 that it is clear that CS ELSI has made major contributions to BBMRI's ELSI activities; further changes to the strategic planning and resource allocation process and the governance structure would improve the efficiency and effectiveness of the service, as well as ensure future sustainability.

Given the thorough analysis and broad consensus within the stakeholders of BBMRI-ERIC on a different approach it is decided not to set up a specific common service on Rare Diseases but rather to incorporate this into the new strategy to develop one central helpdesk, where support for the Rare Disease biobanks can also be given.

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