

## ADOPT BBMRI-ERIC GRANT AGREEMENT NO. 676550

### DELIVERABLE REPORT

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### RD BIOBANK SECTION IN BBMRI-ERIC CATALOGUE

#### Executive Summary

ADOPT Deliverable D7.5 “RD Biobank section in BBMRI-ERIC catalogue” is a software and data deliverable that improves the visibility and accessibility of Rare Disease Biobanks in the BBMRI-ERIC portal. For this we have extended the Directory 3.0 software to create a separate section in the Directory that highlights Rare Disease biobanks, and synergized with efforts in the RD-connect project, the MIABIS standardization effort for minimum information about biobanks and the MOLGENIS open source project. We leveraged the existing data in the BBMRI-ERIC Directory to visualize the potential of this concept and we recognize that as next step a systematic approach to curating and updating the biobank information to list the rare disease biobanks is necessary. This information curation will be part of the operational support of the BBMRI-ERIC directory in the BBMRI-ERIC Common service for IT (CS IT), in collaboration with the BBMRI National Nodes and coordinated by BBMRI-NL.



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## Document log

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## Background

In recent years, biobanks have emerged as an important research resource in both the clinical and translational context. With the advance of medical knowledge, the demand for rare, specialized samples in good condition increases. A growing number of researchers is seeking access to high quality biomaterial outside of their institution. This poses new challenges for the biobanks' information infrastructure, which has to support the new modes of collaboration.

The goal of the ADOPT project is to enable biobanks to combine their efforts and provide a common, highly efficient platform to researchers and further stakeholders. One of ADOPT's deliverables is D7.5 "RD Biobank section in BBMRI-ERIC catalogue". This deliverable addresses the challenge to make rare disease biobanks visible outside of their home institute. The results are described below.

## Approaches (Methods)

The work for this deliverable has been split into two major tasks:

1. Technical realization of a Rare Disease Area in the BBMRI-ERIC Directory [1, 2]
2. Tagging biobank collections as Rare Disease collection

### **Technical realization of the Rare Disease Area in the BBMRI-ERIC Directory**

To create the Rare Disease area in the BBMRI-ERIC Directory we have extended the data model with ability to specify the collection type 'Rare disease Collection' in addition to any of the collection types as defined in the MIABIS [4] Sample Collection module. For this we have largely benefitted from the recent transition of the BBMRI-ERIC Directory from a bespoke web application with an LDAP backend data store to MOLGENIS[3], a flexible web application framework for scientific data that allowed us to rapidly and iteratively develop the new data structures needed. In addition we could capitalize on existing data upload and search capabilities, as well as tools for data harmonization and cleaning and programmatic access (including 'federation' capabilities). On top of this data model we added a dedicated view of the collections to the directory, which pre-filters on the collection type containing the 'Rare disease Collection'. This required that we extended the MOLGENIS software with the functionality to specify queries in the URL and that we could create bookmarkable views [5].

### **Tagging biobank collections as Rare Disease collection**

With the update of the BBMRI-ERIC Directory to version 3, we have reviewed the existing collections and compared them with the listing of Rare Disease biobanks that is publicly available on the RD-Connect ID-Card system [6]. At that point 7 biobanks in the Directory were identified as Rare Disease biobanks and a link to the RD-Connect ID-Card was added for these biobanks. Further the collections of these biobanks have been classified as 'rare disease collection' to make them visible in the dedicated rare disease section that we created as outlined above.

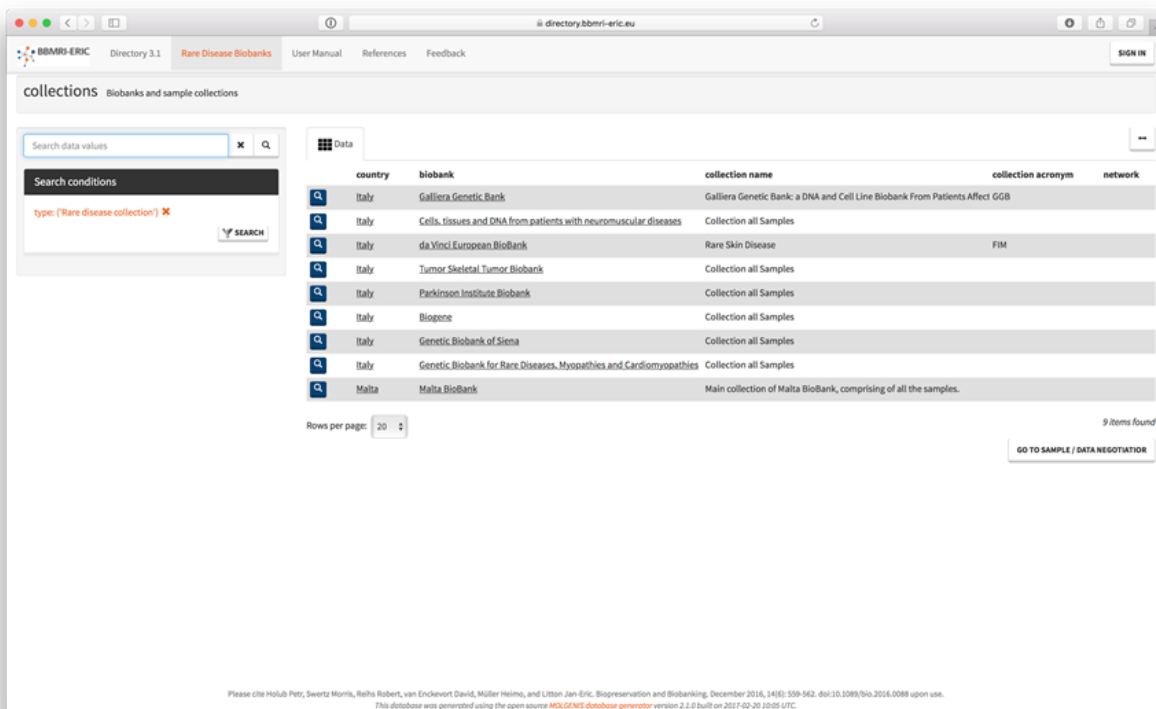
## Schedule

On time



## Results

A new version[7,8] of MOLGENIS was developed, tested and released that supports expressing a search filter in the URL to accommodate the predefined filter for rare disease biobanks. This version has been deployed as the BBMRI-ERIC directory[1] and the Directory has been updated with specific information section on the Rare Disease biobanks.



country	biobank	collection name	collection acronym	network
Italy	Galliera Genetic Bank	Galliera Genetic Bank a DNA and Cell Line Biobank From Patients Affect GGB		
Italy	Cells, tissues and DNA from patients with neuromuscular diseases	Collection all Samples		
Italy	da Vinci European BioBank	Rare Skin Disease	FIM	
Italy	Tumor Skeletal Tumor Biobank	Collection all Samples		
Italy	Parkinson Institute Biobank	Collection all Samples		
Italy	Biogene	Collection all Samples		
Italy	Genetic Biobank of Siena	Collection all Samples		
Italy	Genetic Biobank for Rare Diseases, Myopathies and Cardiomyopathies	Collection all Samples		
Malta	Malta Biobank	Main collection of Malta Biobank, comprising of all the samples.		

Researchers can now easily identify Rare-Disease biobanks by either going to the dedicated section for the Rare Disease biobanks[9] (figure 1) or by using Rare Disease Collection as an additional query on any search in the main page (figure 2).



×
Search biobanks

collections
collection
contact information
donor data
imaging data
sample data
access policy

sample management
data management

collection name

+

collection acronym

+

network

+

type

- Birth cohort
- Case-Control
- Cohort
- Cross-sectional
- Disease specific
- Hospital
- Image collection
- Longitudinal
- Other
- Population-based
- Quality control
- Rare disease collection
- Sample collection
- Twin-study

All software, models and configuration are available as open source at <http://github.com/molgenis> such that national nodes and rare disease networks (such as ERNs) can reuse the system for local cataloguing needs, including a connection to federated data to the central BBMRI-ERIC international biobank directory.

## Discussion and conclusions

The technical implementation of the Rare Disease Section in the Directory was driven by the ADOPT project and could be realized in a short time thanks to the flexibility of the MOLGENIS software. The development of the required functionality in MOLGENIS contributed to new functionality that is available for all MOLGENIS users and also benefitted the development of the integration of the Directory with the Negotiator. As a side benefit a user of the Directory can now also easily bookmark or share a search in the Directory for later use.

The current listing of biobanks in the Rare Disease section of the Directory is incomplete, because the BBMRI-ERIC National Nodes have an incomplete overview of the rare disease biobanks in their country and updates from the National Nodes are only provided periodically.



In order to fully realize the potential of the RD section in the Directory a systematic effort to update the Directory with missing Rare Disease biobank collections needs to be undertaken. This effort should be coordinated with and driven by the BBMRI National Nodes and leverage the existing work in RD-Connect, Orphanet and the Rare Disease ERNs to identify Rare Disease biobanks. Within ADOPT WP7 we have identified The Netherlands and Italy as two National Nodes that have well developed networks of rare disease biobanks and they will function as pilot nodes to make an inventory of the rare disease biobanks in collaboration with the other initiatives. In collaboration with the Dutch representatives of Orphanet the Dutch node is preparing an effort to invite rare disease biobanks to register in the Dutch Node. Other National Nodes should setup similar collaborations to involve and include the rare disease biobanks in the National Node.

To formalize the 'Rare disease collection' classification a formal proposal should be made to the MIABIS Core Working Group in BBMRI-ERIC to consider this classification for inclusion in the MIABIS Core.



## References

- [1] <https://directory.bbmri-eric.eu/>
- [2] Holub Petr, Swertz Morris, Reihls Robert, van Enckevort David, Müller Heimo, and Litton Jan-Eric. Biopreservation and Biobanking. December 2016, 14(6): 559-562. doi:10.1089/bio.2016.0088
- [3] <http://www.molgenis.org>
- [4] Merino-Martinez Roxana, Norlin Loreana, van Enckevort David, Anton Gabriele, Schuffenhauer Simone, Silander Kaisa, Mook Linda, Holub Petr, Bild Raffael, Swertz Morris, and Litton Jan-Eric. Biopreservation and Biobanking. August 2016, 14(4): 298-306. doi:10.1089/bio.2015.0070.
- [5] <https://github.com/molgenis/molgenis/releases/tag/v2.1.0>
- [6] <https://catalogue.rd-connect.eu/>
- [7] MOLGENIS Source Code: <https://github.com/molgenis/molgenis/archive/v2.1.0.zip>
- [8] MOLGENIS Binary Executable (WAR):  
<http://search.maven.org/remotecontent?filepath=org/molgenis/molgenis-app/2.1.0/molgenis-app-2.1.0.war>
- [9] Direct URL to the RD section: [https://directory.bbmri-eric.eu/menu/main/dataexplorer?entity=eu\\_bbmri\\_eric\\_collections&mod=data&attrs%5B%5D=count%5B%5D=biobank&attrs%5B%5D=collection&hideselect=true&filter=type==RD](https://directory.bbmri-eric.eu/menu/main/dataexplorer?entity=eu_bbmri_eric_collections&mod=data&attrs%5B%5D=count%5B%5D=biobank&attrs%5B%5D=collection&hideselect=true&filter=type==RD)

