

# WP3: MAIN ACHIEVEMENT AND CHALLENGES



# RD-Connect tools for biobanks

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## ID-card

The collection of **information on RD-Connect registries and biobanks** to:

- **Search** for registries, biobanks
  - via disease name/ OMIM/ Orphacode
  - filters: registry/biobank
- **Manage** new registry/biobank entries

## Sample Catalogue

The **catalogue of RD-Connect biological samples** to:

- **Search** and request biological samples
  - via disease name/ OMIM/ Orphacode
  - multiple filters: sample material type, availability of genotype data, sex, etc
- **Manage** sample workflow



# Achievement: sample catalogue

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- Sample catalogue is available for testing at:  
<https://molgenis10.target.rug.nl/>
  - Proof-of-concept: Ring chromosome 14 biobank/registry
- We are now populating the sample catalogue with samples from the following biobanks:
  - TNGB (Telethon Network of Genetic Biobanks: 11 Italian biobanks, ongoing)
  - Bank of the Molecular Genetics Laboratory, *Universita ta Malta* (ongoing)
  - Bank of the Research Institute for Rare Diseases, *Instituto de Salud Carlos III*, Madrid (starting now)
  - MRC Centre for Neuromuscular Diseases BioBank, University of Newcastle (planned)



# Achievement: sample catalogue & ID-CARD disease matrix

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## SAMPLE CATALOGUE → ID-CARD

1. Each sample in the sample catalogue is linked to the ID-Card of the hosting biobank
2. (Where available) the sample is also linked to the ID-Card of the registry collecting the clinical data of the patient

## ID-CARD → SAMPLE CATALOGUE

1. The disease matrix for each biobank is created by aggregating the data in the sample catalogue by disease
2. Proof-of-concept: by clicking on a specific disease in the disease matrix of a biobank, the user is re-directed to a filtered page of the sample catalogue listing all the samples for that disease available in that biobank

# Sample Information

Search data values



## Data item filters

Material Type: ('DNA') ✕

Disease: ('Ring chromosome 14') ✕

Wizard

## Data item selection

Select all Deselect all

- Sample ID
- MIABIS Material Type
- Material Type
- Anatomical Site
- Year of Sampling
- Sample Collection
- Hosting Biobank
- Disease
- Year of Birth
- Year of Death
- Year of Diagnosis



Data



Aggregates



Charts



	MIABIS Material Type	Material Type	Anatomical Site	Hosting Biobank	Disease
	DNA	DNA	Blood	Galliera Genetic Bank	Ring chromosome 1
	DNA	DNA	Blood	Galliera Genetic Bank	Ring chromosome 1
	DNA	DNA	Blood	Galliera Genetic Bank	Ring chromosome 1
	DNA	DNA	Blood	Galliera Genetic Bank	Ring chromosome 1
	DNA	DNA	Blood	Galliera Genetic Bank	Ring chromosome 1
	DNA	DNA	Blood	Galliera Genetic Bank	Ring chromosome 1
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	DNA	DNA	Blood	Galliera Genetic Bank	Ring chromosome 1
	DNA	DNA	Blood	Galliera Genetic Bank	Ring chromosome 1
	DNA	DNA	Blood	Galliera Genetic Bank	Ring chromosome 1
	DNA	DNA	Blood	Galliera Genetic Bank	Ring chromosome 1





# Link to the registry

RD-Connect

Sample Catalogue

Biobanks ▾

Account

## RD-Connect Sample Catalogue

Search data values



### Data item filters

Wizard

### Data item selection

Select all Deselect all

- Sample ID
- Material Type
- Anatomical Site
- Sex
- Disease
- Diagnosis Type
- Age at Sampling
- Age at Death
- Age at Diagnosis
- Age at Remission
- Affected
- Family members available
- Related samples available
- Genotype data available
- Registry

Data Aggregates Charts

<input checked="" type="checkbox"/> Material Type	<input checked="" type="checkbox"/> Anatomical Site	<input checked="" type="checkbox"/> Disease	<input checked="" type="checkbox"/> Diagnosis Type	<input checked="" type="checkbox"/> ID-CARD
DNA	Blood	Ring chromosome 14	Molecular,Cytogenetics	Ring14 Clinical database
Leukocyte	Blood	Ring chromosome 14	Molecular,Cytogenetics	Ring14 Clinical database
Leukocyte	Blood	Ring chromosome 14	Molecular,Cytogenetics	Ring14 Clinical database
Lymphoblast	Blood	Ring chromosome 14	Molecular,Cytogenetics	Ring14 Clinical database
Leukocyte	Blood	Ring chromosome 14	Molecular,Cytogenetics	Ring14 Clinical database
DNA	Blood	Ring chromosome 14	Molecular,Cytogenetics	Ring14 Clinical database
Leukocyte	Blood	Ring chromosome 14	Molecular,Cytogenetics	Ring14 Clinical database
Fibroblast	Muscle tissue	Ring chromosome 14	Molecular,Cytogenetics	Ring14 Clinical database
DNA	Blood	Ring chromosome 14	Cytogenetics	Ring14 Clinical database
Leukocyte	Blood	Ring chromosome 14	Molecular,Cytogenetics	Ring14 Clinical database
Lymphoblast	Blood	Ring chromosome 14	Molecular,Cytogenetics	Ring14 Clinical database
DNA	Blood	Ring chromosome 14	Molecular,Cytogenetics	Ring14 Clinical database
Lymphoblast	Blood	Ring chromosome 14	Molecular,Cytogenetics	Ring14 Clinical database
DNA	Blood	Ring chromosome 14	Molecular,Cytogenetics	Ring14 Clinical database
Lymphoblast	Blood	Ring chromosome 14	Molecular,Cytogenetics	Ring14 Clinical database
DNA	Blood	Ring chromosome 14	Molecular,Cytogenetics	Ring14 Clinical database
Lymphoblast	Blood	Ring chromosome 14	Molecular,Cytogenetics	Ring14 Clinical database
DNA	Blood	Ring chromosome 14	Molecular,Cytogenetics	Ring14 Clinical database

ID-CARD

Type	Target population	Website	ID-Card
registry	International	<a href="http://www.ring14.org/eng/120/database/">http://www.ring14.org/eng/120/database/</a>	<a href="http://catalog">http://catalog</a>

Rows per page: 20

1 item found

Ok



# Challenges: sample loading

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Open Issues	Identified solutions
a) The <b>data model</b> of each biobank may be different from the RD-Connect data model	A <b>mapping service</b> module to facilitate the mapping of multiple datasets to the catalogue data model was developed (BiobankConnect)
b) To develop <b>automated systems for the update</b> of RD-Connect catalogue after the first massive uploading of samples	<b>Two models</b> of data updating/loading are under development: <ul style="list-style-type: none"><li>▪ «<b>push</b>» model where the data are provided by the biobank system to the RD-Connect sample catalogue</li><li>▪ «<b>pull</b>» model where the RD-Connect catalogue itself retrieves the data from the biobank's database</li></ul>



# Progress in full workflow for sample request

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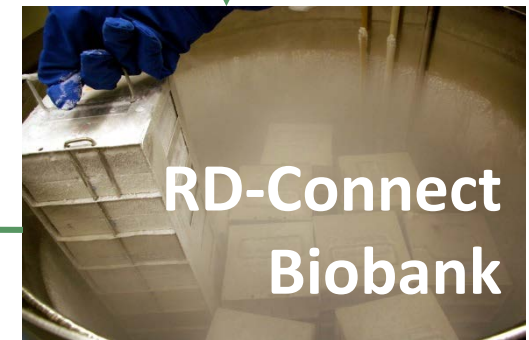
Donor/Clinician/Investigator

provides a signed

**Informed Consent**

**Sample**

**Sample Deposit form**



User  
**User registration form**

**MTA**  
**Cost Recovery form**  
**Sample shipping form**  
**Sample receipt confirmation**

 **Sample Catalogue**  
RD-Connect integrated platform

**Sample request form**

**Sample use confirmation**  
**Feedback questionnaire**  
**Scientific Report**

**Implement all forms into the user interface**





# Progress in including new biobanks

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- Achievements
  - Request/assessment workflow completed
  - Assessment panel established (from among EBB members)
- Next step
  - Implement the process





# Progress in integrating EuroBioBank into RD-Connect

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## □ Achievements

- Integration of EBB into RD-Connect approved by the EMC and by the EBB Assembly in 2015
- 13<sup>th</sup> EBB Assembly meeting back-to-back the 2016 RD-Connect meeting
- 14 biobanks in the process of uploading their sample data in the RD-Connect sample catalogue
- Participation in the RD-Connect biobank Assessment Panel



- **Established in 2001, the first RD biobank network in Europe**
- **26 Members, 23 Biobanks in 11 Countries**
- **150 k samples stored, >950 rare diseases**
- **Biobanking SOPs**

## □ Next steps

- Ensure visibility in the RD-Connect platform
- Contribute to SOPs and ELSI management
- Merging/integrating EBB charter into RD-Connect code of practice



# Synergy with BBMRI-ERIC

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- Achievements
  - Preparatory meeting in Milan on the 13th of May 2015 (UNEW, FTELE, BBMRI-ERIC)
  - BBMRI-ERIC full partner in RD-Connect (end 2015)
  - Main synergic activities
    - Biobank assessment
    - Sample catalogue
    - ELSI issues
    - Training activities
    - Sustainability prospect
- Next step
  - Establish working group to build on existing synergies



# WP3 summary

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- Main Achievements
  - The sample catalogue is running (beta version)
  - The sample catalogue is integrated with ID-Card
  - The workflows for sample management and new biobank integration are completed
- Challenges/next steps
  - To populate the RD-Connect sample catalogue with biobanks' sample data
  - To implement the full workflow for sample management and for including new biobanks
  - To integrate EuroBioBank into RD-Connect
  - To build on synergies with BBMRI-ERIC



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