

# BROWSING SAMPLE COLLECTIONS IN SAMPLE CATALOGUE

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# Acknowledgements/collaborations

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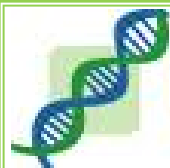


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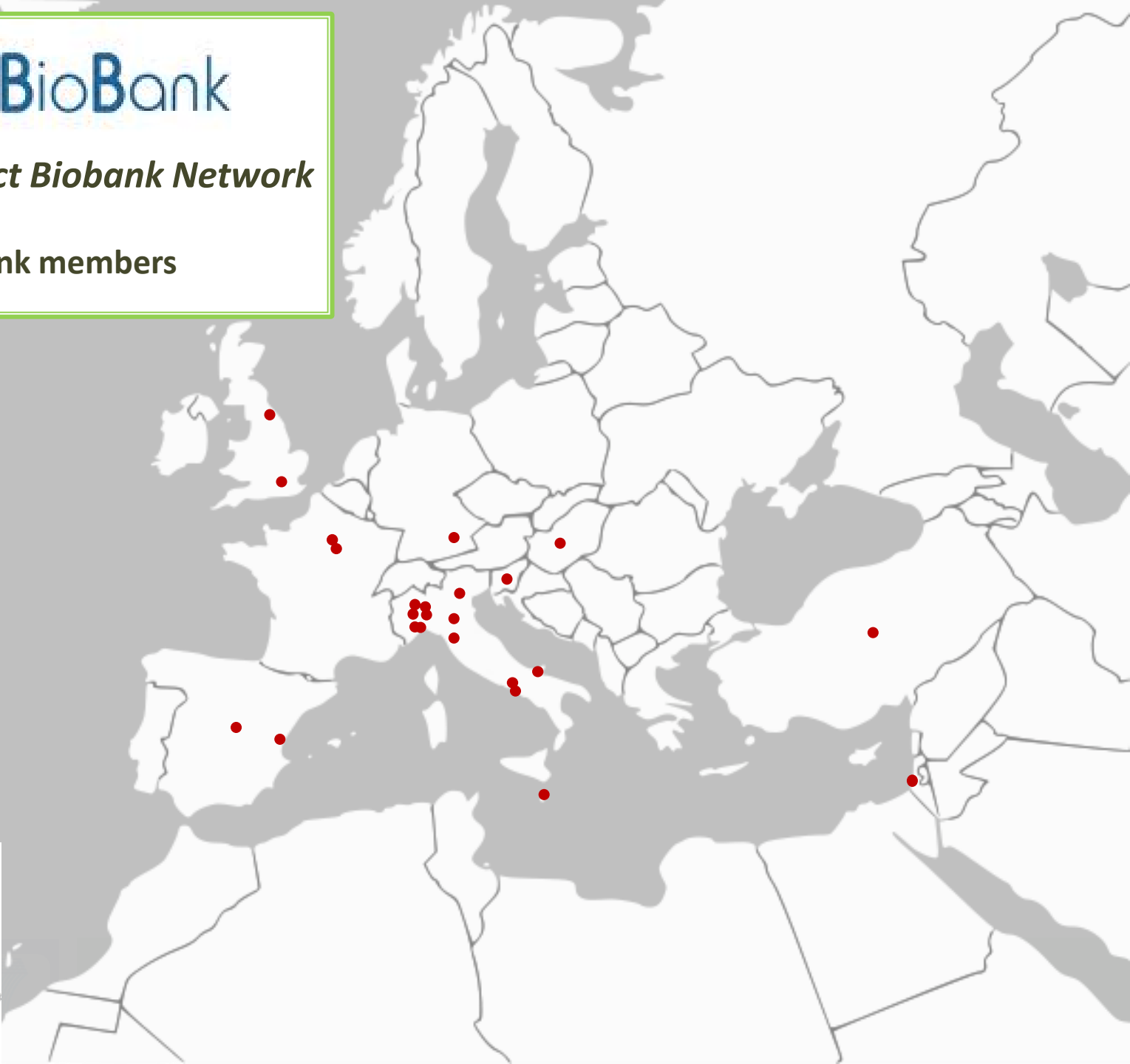
RD-Connect Consortium



# EuroBioBank

*The RD-Connect Biobank Network*

● 25 RD biobank members





# Flagship tools for RD research



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## RD Connect

An integrated platform for rare disease research

**SAMPLE CATALOGUE**



**7352** BIOSAMPLES COVERING  
**90** RARE DISEASES

**REGISTRY & BIOBANK  
FINDER**



**360** RARE DISEASE PATIENT  
REGISTRIES AND BIOBANKS

**GENOME-PHENOME  
ANALYSIS PLATFORM**



**2605** GENOMES & EXOMES  
OF PATIENTS AND FAMILIES



# The tools for biobanks



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	Registry & Biobank Finder	Sample Catalogue
<i>What is it?</i>	A collection of information on existing rare disease registries and biobanks	A centralised catalogue of rare disease biological samples
<i>What can you do with it?</i>	Search for registries, biobanks	Search (and request) for biological samples
<i>Search via?</i>	Disease name, OMIM, Orphacode Filters: Registry/disease	Disease name, OMIM, Orphacode Multiple filters: sample material type, availability of genotype data, sex, etc
<i>What else?</i>	Management of new registry/biobank entries	Management of sample workflow ( <i>underdevelopment</i> )



# RD-Connect Sample Catalogue



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RD  Connect

SAMPLE



CATALOGUE

<https://samples.rd-connect.eu/>



# Use case: searching for samples



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**Ring chromosome 14 syndrome** is a rare disorder that is characterized by abnormalities of the 14th chromosome. It is characterized by early-onset epilepsy, developmental delay with mental retardation, microcephaly, and dysmorphic facial features.

“I want to find **10 DNA samples** from **females** with **Ring Chromosome 14 Syndrome**”.

*image from Knijnenburg et al., 2007 EJHG*



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## Search

Disease name  
“ring chromosome 14”

Or

Orpha number  
“1440”

Search input field (highlighted in yellow)

Data item filters

Wizard

Data item selection

Select all Deselect all

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

Data	Aggregates
Disease	
Duchenne muscular dystrophy	
Duchenne muscular dystrophy	
Duchenne muscular dystrophy	
Duchenne muscular dystrophy	
Duchenne muscular dystrophy	
Duchenne muscular dystrophy	
Becker muscular dystrophy	
Becker muscular dystrophy	
Myofibrillar myopathy	
Autosomal recessive limb-girdle muscular dystrophy type 2A	
Duchenne muscular dystrophy	
Duchenne muscular dystrophy	
Duchenne muscular dystrophy	
Duchenne muscular dystrophy	
Duchenne muscular dystrophy	
Duchenne muscular dystrophy	
Becker muscular dystrophy	
Becker muscular dystrophy	
Duchenne muscular dystrophy	



1 ring chromosome 14 x

2 Data Aggregates

3

4

5

Data item filters

Wizard

Data item selection

Select all Deselect all

☒ Sample ID

☒ Disease

☒ Material Type

☒ Anatomical Site

☒ Sex

☒ Diagnosis Type

☒ Genotype data available

☒ Age at Sampling

☒ Age at Death

☒ Age at Diagnosis

☒ Age at Remission

☒ Affected

☒ Family members available

☒ Related samples available

☒ Registry data available

☒ Hosting Biobank

☒ Hosting Registry

☒ Participant ID

Filter

Disease	Material Type	Anatomical Site	Sex	Diagnosis Type	Genotype
Ring chromosome 14	DNA	Blood	Male	Molecular,Cytogenetics	Yes
Ring chromosome 14	Leukocyte	Blood	Female	Molecular,Cytogenetics	Yes
Ring chromosome 14	Lymphoblast	Blood	Female	Molecular,Cytogenetics	Yes
Ring chromosome 14	Lymphoblast	Blood	Female	Molecular,Cytogenetics	Yes
Ring chromosome 14	DNA	Blood	Female	Molecular,Cytogenetics	Yes
Ring chromosome 14	Leukocyte	Blood	Male	Molecular,Cytogenetics	Yes
Ring chromosome 14	Lymphoblast	Blood	Male	Molecular,Cytogenetics	Yes
Ring chromosome 14	DNA	Blood	Male	Molecular,Cytogenetics	Yes
Ring chromosome 14	Leukocyte	Blood	Male	Molecular,Cytogenetics	Yes
Ring chromosome 14	Lymphoblast	Blood	Male	Molecular,Cytogenetics	Yes
Ring chromosome 14	DNA	Blood	Female	Molecular,Cytogenetics	Yes
Ring chromosome 14	Leukocyte	Blood	Female	Molecular,Cytogenetics	Yes
Ring chromosome 14	Lymphoblast	Blood	Female	Molecular,Cytogenetics	Yes
Ring chromosome 14	Lymphoblast	Blood	Female	Cytogenetics	Yes
Ring chromosome 14	Lymphoblast	Blood	Female	Cytogenetics	Yes
Ring chromosome 14	Leukocyte	Blood	Male	Molecular,Cytogenetics	Yes
Ring chromosome 14	Lymphoblast	Blood	Male	Molecular,Cytogenetics	Yes
Ring chromosome 14	DNA	Blood	Male	Molecular,Cytogenetics	Yes
Ring chromosome 14	Lymphoblast	Blood	Female	Molecular,Cytogenetics	Yes
Ring chromosome 14	DNA	Blood	Female	Molecular,Cytogenetics	Yes

Rows per page:

Previous 1 2 3 4 5 Next

6 98 items found

"I want to find **10 DNA samples** from **females** with **Ring Chromosome 14 Syndrome**".



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ring chromosome 14

**Data item filters**

**Data item selection**

Select all Deselect all

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



ring chromosome 14

**Data item filters**

Sex.Preferred term: female

Material Type.Preferred term: dna

**Data item selection**

Select all Deselect all

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

**Filter**

Material type  
"DNA"

Sex  
"Female"



“I want to find **10 DNA samples from females with Ring Chromosome 14 Syndrome**”.



1440 ✕ 🔍

**Data item filters**

Sex.Preferred term: female ✕

Material Type.Preferred term: dna ✕

🔍 Wizard

**Data item selection**

Select all Deselect all

☐ 🔍 Sample ID

☒ 📄 Disease

☒ 📄 Material Type

☐ 📄 Anatomical Site

☒ 📄 Sex

☐ 📄 Diagnosis Type

☐ 📄 Genotype data available

☐ 🔍 Age at Sampling

☐ 🔍 Age at Death

☐ 🔍 Age at Diagnosis

☐ 🔍 Age at Remission

📊 Data 📈 Aggregates ↔

<input checked="" type="checkbox"/> Disease	<input checked="" type="checkbox"/> Material Type	<input checked="" type="checkbox"/> Sex
Ring chromosome 14	DNA	Female
Ring chromosome 14	DNA	Female
Ring chromosome 14	DNA	Female
Ring chromosome 14	DNA	Female
Ring chromosome 14	DNA	Female
Ring chromosome 14	DNA	Female
Ring chromosome 14	DNA	Female
Ring chromosome 14	DNA	Female
Ring chromosome 14	DNA	Female
Ring chromosome 14	DNA	Female
Ring chromosome 14	DNA	Female
Ring chromosome 14	DNA	Female
Ring chromosome 14	DNA	Female
Ring chromosome 14	DNA	Female

**Rows per page:** 14 items found



# Locate the biological samples



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“I want to know which **biobanks** hold **plasma** from **females** with **Steinert myotonic dystrophy**.”

Data item filters

Sex.Preferred term: female

Material Type.Preferred term: plasma

Data item selection

☐  Sample ID

☒  Disease

☐  MIABIS Material Type

☒  Material Type

☐  Anatomical Site

☒  Sex

Group by

Hosting Biobank

x

Material Type

Distinct

Select ...

	Plasma	Total
Galliera Genetic Bank	35	35
Newcastle MRC Biobank for Rare and Neuromuscular Diseases	7	7
Total	42	42



## RD-Connect Sample Catalogue

- A tool for rare disease biobanks to make their sample collections findable and accessible.
- Allows researchers to easily search for biological samples via diseases names, material types, and many other filters.
- Allows researchers to identify and locate the biobanks holding the biological samples.
- Open for all users.
- More information: <http://rd-connect.eu/>