

Finding registries and biosamples  
in just few clicks:  
**Registry & Biobank Finder and  
Sample Catalogue**

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# Flagship tools for RD research

## RD Connect

An integrated platform for rare disease research

### SAMPLE CATALOGUE



**22,932** BIOSAMPLES  
COVERING **212** RDS

### REGISTRY & BIOBANK FINDER



**382** RD PATIENT REGISTRIES  
AND BIOBANKS

### GENOME-PHENOME ANALYSIS PLATFORM



**3,894** GENOMES & EXOMES  
OF PATIENTS AND FAMILIES



# Definitions

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## □ What is a **registry**?

... a patient registry is an *organized system* that uses observational study methods to collect uniform data (clinical and other) to evaluate specified outcomes for a population defined by a particular disease, condition, or exposure, and that serves one or more predetermined scientific, clinical, or policy purposes.

(Agency for Healthcare Research and Quality, AHRQ)

## □ What is a **biobank**?

Biobank is a biorepository that accepts, processes, stores and distributes biospecimens and associated data for use in research and clinical care

(De Souza and Greenspan, 2013 *Biobanking Past, Present and Future: Responsibilities and Benefits*)



# Sample Catalogue

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

Sample Catalogue contains information on available samples across participating biobanks

22,932 samples covering 10 material types, representing more than 200 rare diseases.



# Use case: searching for samples

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**Duchenne muscular dystrophy** (DMD) is a genetic disorder characterized by progressive muscle degeneration and weakness. Affected persons develop problems walking and breathing. It is caused by mutation in *dystrophin* gene, which encodes for protein that anchors cytoskeleton of a muscle fiber to the surrounding extracellular matrix.

DMD is a X-linked recessive disorder.

“I want to find **2 myoblasts samples** from **females** with **Duchenne Muscular Dystrophy**”.





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## Search

Disease name

**Duchenne Muscular dystrophy**  
**DMD**

Or

Orpha number  
**98896**

OMIM not available

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
Becker muscular dystrophy

## Filter

I want to find 2 myoblasts samples from **females** with **Duchenne Muscular Dystrophy**



# Locate the biological samples

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“I want to know which **biobank(s)** hold **samples** from patients with **Congenital muscular dystrophy type 1A.**”

**MCD1A**, muscular dystrophy caused by mutations in the *LAMA2* gene and is inherited in an autosomal recessive manner.

Congenital muscularular ✕ 🔍

Search criteria

Set up

Data item selection

Select all Deselect all

☒ Disease

☒ Material Type

☒ Anatomical Site

☒ Sex

☒ Diagnosis Type

☒ Genotype data available

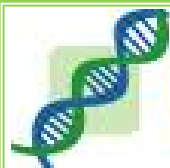
☐ Age at Sampling

Data Aggregates

Group by Material Type x Hosting Biobank

Distinct Select ...

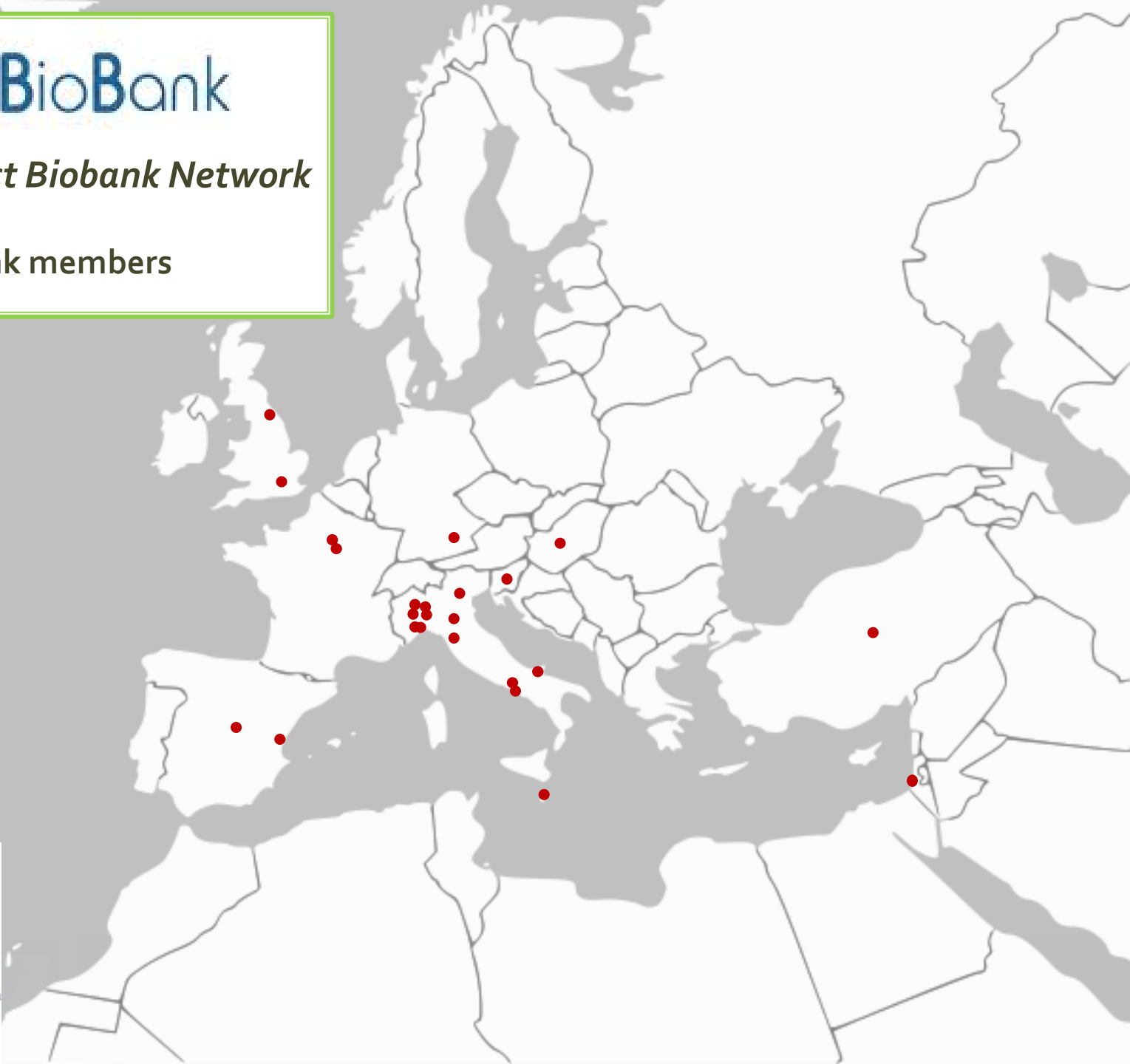
	Biobank of the Institute of Rare Diseases Research/Institute of Health Carlos III (IIR-ISCIII)	Newcastle MRC Biobank for Rare and Neuromuscular Diseases	Total
Leukocyte	1	0	1
Portion of plasma	1	0	1
Portion of serum	1	4	5
DNA	1	0	1
Total	4	4	8



# EuroBioBank

*The RD-Connect Biobank Network*

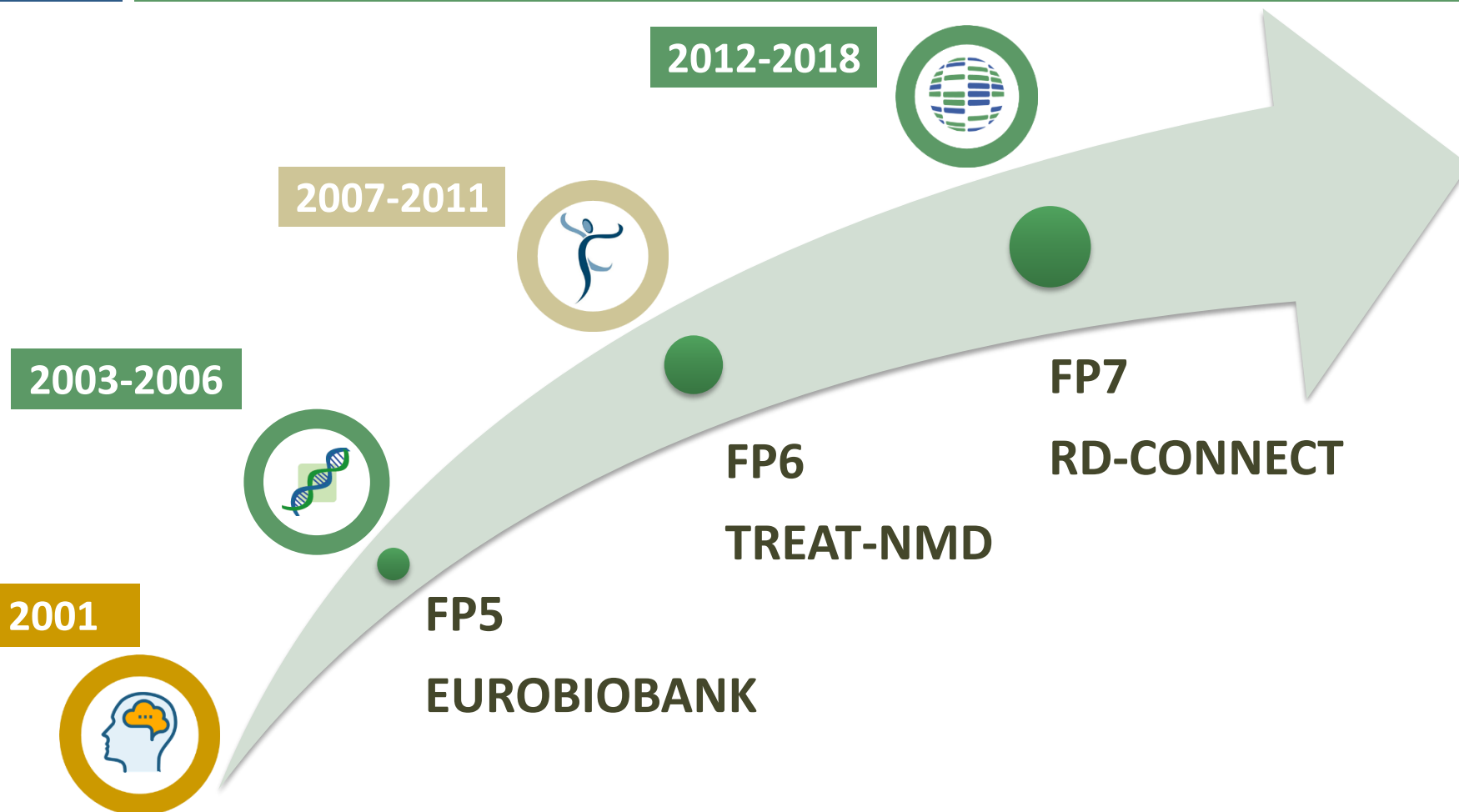
● 25 RD biobank members





# Interest and investment from EC

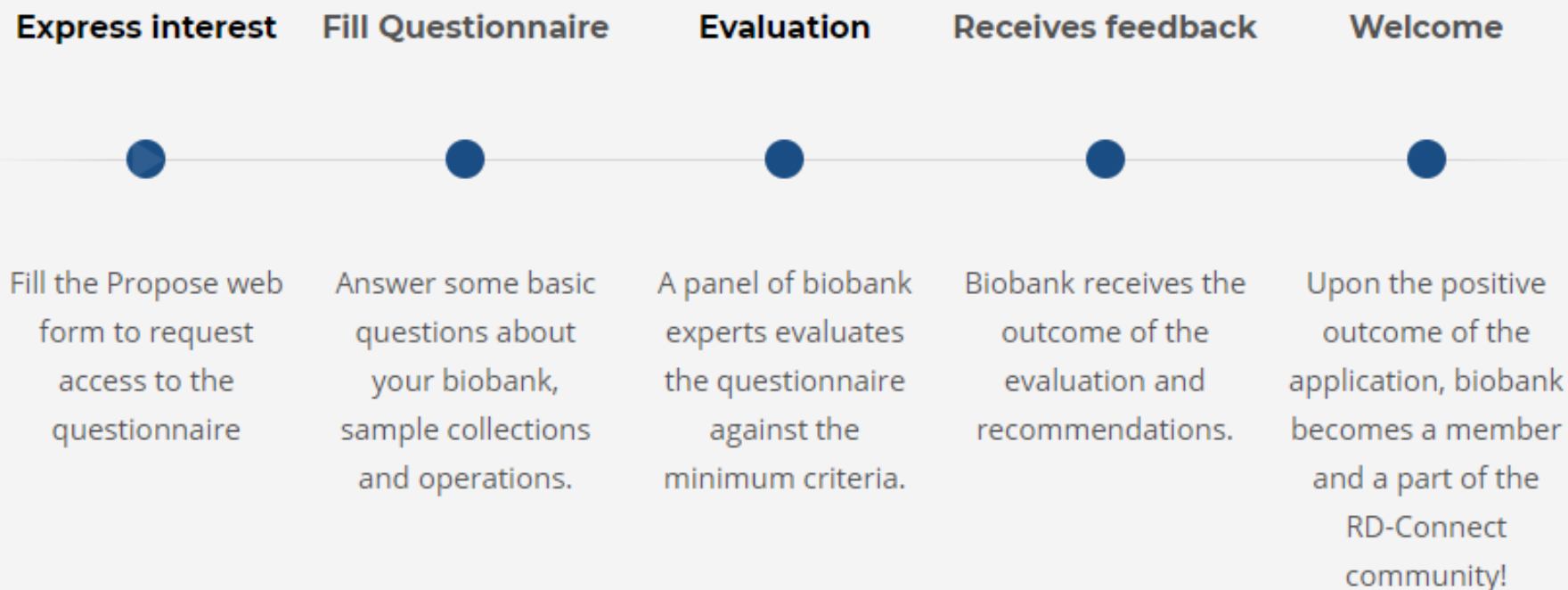
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# Joining EuroBioBank

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Based on biobank self-declaration.

More info <https://rd-connect.eu/biosamples-data/biobank-assessment-process/>



# Registry & Biobank Finder

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

Global directory of RD patient registries and peer-reviewed biobanks. Displays basic information on contacts persons, collections, as well as some study documents or SOPs.

Currently it lists 360 patient registries and 22 biobanks, covering over 1500 RDs.



# Resources

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Gainotti et al., 2018. <https://doi.org/10.1038/s41431-017-0085-z>

European Journal of Human Genetics (2018) 26:631–643  
<https://doi.org/10.1038/s41431-017-0085-z>



## ARTICLE



### The RD-Connect Registry & Biobank Finder: a tool for sharing aggregated data and metadata among rare disease researchers

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How to navigate the RD-Connect Registry & Biobank Finder

<https://youtu.be/ooMAXGFdseI>

How to add a registry or biobank to the RD-Connect Registry & Biobank Finder

<https://youtu.be/uDSSwftYvsk>



# Summary

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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, browse for biological samples
<i>Search via?</i>	Disease name, OMIM, Orphacode Filters: Registry/disease	Disease name, Orphacode Multiple filters: sample material type, availability of genotype data, sex, etc
<i>What else?</i>	Management of new registry/biobank entries	Request of samples ( <i>via BBMRI-ERIC Negotiator, underdevelopment</i> )