

Guide to the RD-Connect Registry & Biobank Finder

for managers of biobanks and patient registries



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What is the RD-Connect Registry & Biobank Finder?

Registry & Biobank Finder is a comprehensive and searchable global directory of rare disease biobanks and patient registries, created by the [RD-Connect project](#).

The purpose of biobanks and registries is to collect and share biosamples and clinical information from rare disease patients, but it is often difficult for researchers to find out what each resource contains and how to access it. Registry & Biobank Finder allows finding biobanks and registries with data and samples on a particular rare disease, including the numbers of cases and contact information.

Why sign up to the RD-Connect Registry & Biobank Finder directory?

Joining Registry & Biobank Finder is an excellent way to increase the impact of your biobank/registry and to encourage researchers and the rare disease community to use your collections.

Being part of Registry & Biobank Finder:

- lets researchers see a detailed overview of what samples/data you have and how to contact you
- helps you get more involved in research and innovative data-sharing mechanisms
- keeps your data up-to-date in other catalogues like the Orphanet system.

In the system, each biobank/registry has its own “ID-card” that provides a summary of the diseases you cover, the number of cases, and other important information about your resource.

Each ID-card is organised into five sections:

- 1. General overview and organisation**
- 2. Diseases collected**
- 3. Standards and procedures**
- 4. Accessibility of data**
- 5. Study documents e.g. study protocol, case report form, informed consent template and data access agreement.**

Getting started

Visit <http://catalogue.rd-connect.eu/> to access the Registry & Biobank Finder homepage. If you are new to the RD-Connect online catalogue, select the **Propose** button to get started (marked with the red arrow).



To create an ID card for your biobank/registry you need to enter data for your basic profile:

Country	<i>Enter the country where biobank/registry is based.</i>
Type	<i>Select one of: Registry, Biobank, Biobank/registry – select registry if you only hold data and no samples, biobank if you are primarily a resource for sample banking and distribution, and biobank/registry if you not only hold samples but also clinical data about the patient the sample came from.</i>
Acronym	<i>If your resource is known by a project acronym or other short name, enter it here.</i>
Name	<i>Name of Biobank/registry - Mandatory Field</i>
Address	<i>Enter the address that someone wanting to contact you should use - Mandatory Field</i>
Contact Person/ Email	<i>Main point of contact for enquiries via catalogue - Mandatory Field</i>
URL	<i>Biobank/registry website address</i>
Head of Registry/ Biobank	<i>If different from person creating catalogue profile</i>
Disease Codes	<i>If possible enter at least one disease code for each disease your resource deals with. You can enter multiple disease codes separated by a semicolon (;) and can use ORPHA number, ICD10, OMIM, UMLS, MESH, MEDDRA, SNOMEDCT)</i>
Disease Names	<i>You can enter multiple diseases separated by a semicolon (;) – if possible please use the disease name as it appears in the Orphanet Rare Disease Ontology.</i>

RD Connect already a member? [Sign In](#)

Propose Candidate

Country: Type: Acronym:

Name: Contact Person:

Address: Contact E-Mail:

URL: Head of Registry/Biobank:

Disease Codes: Comment:

Disease Names:

Once you select **Propose** you will have completed your initial registration. You will then receive an email from catalogue@rd-connect.eu containing your username and password. This will allow you to log in. Follow the hyperlink within the email in order to log in for the first time. At this stage you will also be asked to change your password to one you set for yourself. If you do not receive your log in details within 24 hours, please contact rd-connect.wp2@iss.it to request them.

When logging in from the [Registry & Biobank Finder homepage](#), use the **Sign in** button located in the top right hand side of the screen. Upon logging in to the catalogue for the first time you are taken to your **Overview** section, where you are able to see all of the basic information about your biobank/registry that was entered upon registration (as shown below).

RD Connect my organisations

GNEM-DMP Date of Inclusion: 15/12/2015 Last Activity: 05/12/2015

Overview Diseases Standards Accessibility Documents

General Information

GNE Myopathy Disease Monitoring Program (GNEM-DMP)

Type of Host Institution
Source of funding
Target population
Year of establishment
Ontologies used
Imaging available
Also listed in:

Personnel


Main contact

Philip Cammish
phillip.cammish@ncl.ac.uk

Staff [Add One](#)

Editing Your Biobank/Registry Information

The next stage of the process is to add further relevant information about your biobank/registry.

The **edit buttons**  on the Overview page let you edit and provide additional information to be displayed. The following categories can be edited:

Edit Organization

Organization Information:

Change the biobank/registry's name in the **Details** area and add your logo (as shown on the picture).

Categorization:

Add Tags (words or phrases) that will make your biobank/ registry easier to find via the "Search" tool (similar to keywords that are used on websites). You can add as many tags as you like.

Identification:

In this area, add your address, phone number(s), additional email addresses, website links as well as services (opening times). This information will appear on your **Overview** page.

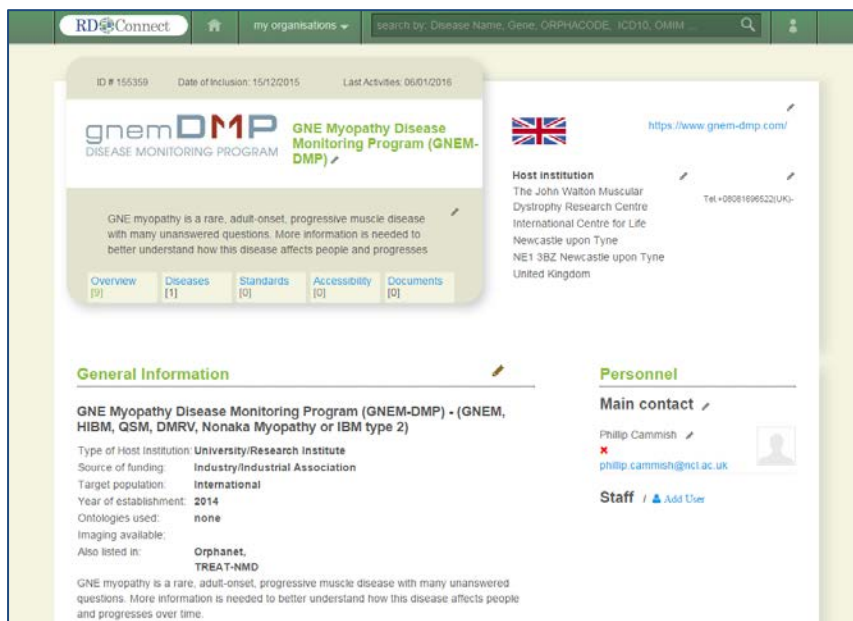
Edit Core

Description - Add information about your biobank /registry that will appear under the general information section on your homepage. You can use editing tools to alter the style and format of text in this area. You can also add **Acronyms** if appropriate that will appear next to the biobank/registry title.

Within the **Edit Core** section, you can also enter further information if applicable. It will appear under the biobank/registry title on your homepage:

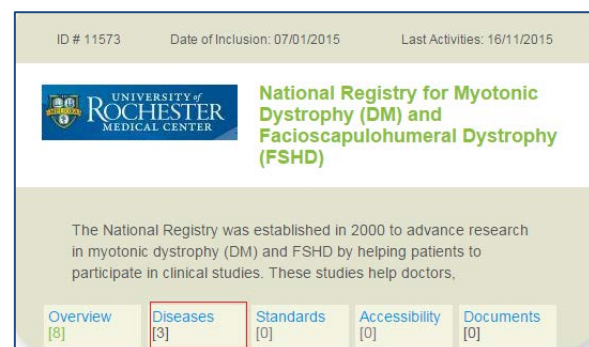
- **Type of host institution**
- **Source of funding**
- **Target Country**
- **Target population of the registry**
- **Year of establishment**
- **Ontologies**
- **Additional Ontologies**
- **Associated data available**
- **Additional Associated data available**
- **Imaging available**
- **Additional Imaging available**
- **The registry is listed in other inventories/networks**
- **Additional networks/inventories**

A number of options shown above let you select multiple options from the pre-populated list. To do this, hold the **Ctrl** button on your keyboard and use your mouse cursor to select the multiple options. Once you have finished the amendments to the **Edit Core** section, select **Save**. Your Overview page will look similar to the one shown below:



Diseases

Within this area you can edit the **Disease Matrix**. The Disease Name, Number of Patients/Donors, Gene, ORPHA Code, ICD10, OMIM, Synonym(s) can all be added to this area. If the registry or biobank that you are editing has more than one disease associated with it, you are able to add multiple entries using the **Add Disease** button.



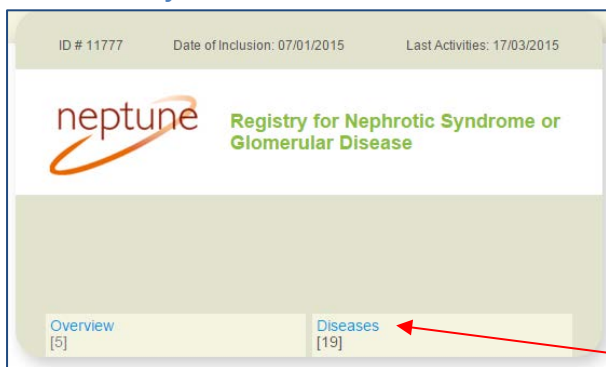
Disease Matrix

Add Disease Scegli file Nessun file selezionato Save Download Disease Matrix as XLS File
Download Disease Matrix as XLSX File

Disease Name	Number of Patients, Donors	Gene	ORPHA Code	ICD 10	OMIM	Synonym(s)	Modified Date	
Nephrotic syndrome, type 2	null	PDCN			#600995		06.03.2014 09:40:45	▼ Actions
Nephrotic syndrome, type 3	null	PLCE1			#610725		06.03.2014 09:42:33	▼ Actions
Nephrotic syndrome, type 4	null	WT1			#256370		06.03.2014 09:43:47	▼ Actions
Nephrotic syndrome, type 1	null	NPHS1	ORPHA839	N04	#256300		14.03.2014 14:06:00	▼ Actions
Nephrotic syndrome, type 8	null	ARHGD1A			#615244		06.03.2014 09:58:18	▼ Actions

Within the **Disease Matrix** section you can also import/export data, e.g. export the matrix in an excel file, where you can change existing disease information and import it back to the system (using the **Choose File** button and then **Save**). A **Modified Date** section appears on the right hand side of the matrix and will update each time the information for each disease has been edited.

How to edit your disease matrix - method 1



Click "Diseases"

Disease Matrix

Nephrotic syndrome, type 2

Disease Name
Nephrotic syndrome, type 2

Number of Patients, Donors
null

Gene
PDCN

ORPHA Code

ICD 10

OMIM
#600995

Synonym(s)

Save Cancel

Disease Matrix

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Download Disease Matrix as XLSX File

Disease Name	Number of Patients, Donors	Gene	ORPHA Code	ICD 10	OMIM	Synonym(s)	Modified Date	
Nephrotic syndrome, type 2	null	PDCN			#600995			▼ Edit X Delete
Nephrotic syndrome, type 3	null	PLCE1			#610725		09:42:33	▼ Actions
Nephrotic syndrome, type 4	null	WT1			#256370		06.03.2014 09:43:47	▼ Actions
Nephrotic syndrome, type 1	null	NPHS1	ORPHA839	N04	#256300		14.03.2014 14:06:00	▼ Actions
Nephrotic syndrome, type 8	null	ARHGD1A			#615244		06.03.2014 09:58:18	▼ Actions

Select "Actions" and then "Edit"

Add the number of patients/donors with the diagnosis and click "Save"

How to edit your disease matrix - method 2

Disease Matrix

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[Download Disease Matrix as XLSX File](#)

Disease Name	Number of Patients, Donors	Gene	ORPHA Code	ICD 10	OMIM	Synonym(s)	Modified Date	Actions
Nephrotic syndrome, type 2	null	PDCN			#600995		06.03.2014 09:40:45	Actions
Nephrotic syndrome, type 3	null	PLCE1			#610725		06.03.2014 09:42:33	Actions
Nephrotic syndrome, type 4	null	WT1			#256370		06.03.2014 09:43:47	Actions
Nephrotic syndrome, type 1	null	NPHS1	ORPHA839	N04	#256300		14.03.2014 14:06:00	Actions
Nephrotic syndrome, type 8	null	ARHGD1A			#615244		06.03.2014 09:58:18	Actions

download the disease matrix as xls file

A1 diseasematrix.xls diseasematrix.xls

A	B	C	D	E	F	G	H
diseasematrixid	diseasename	patientcount	gene	orpha	icd10	omim	sy
107	Nephrotic syndrome, type 2	null	PDCN			#600995	
109	Nephrotic syndrome, type 3	null	PLCE1			#610725	
116	Nephrotic syndrome, type 4	null	WT1			#256370	
131	Nephrotic syndrome, type 1	null	NPHS1	ORPHA83	N04	#256300	
134	Nephrotic syndrome, type 8	null	ARHGD1A			#615244	
140	Nephrotic syndrome, type 9	null	ADCK4			#615573	
149	Nephrotic syndrome, type 5, with or without ocular abnormalities	null	LAMB2			#614199	
153	Nephrotic syndrome, type 6	null	PTPRO			#614196	
156	Nephrotic syndrome, type 7	null	DGKE			#615008	
166	Glomerulosclerosis, focal segmental, 4, susceptibility to	null	APOL1			#612551	
169	Glomerulosclerosis, focal segmental, 3	null	CD2AP			#607832	
178	Glomerulosclerosis, focal segmental, 6	null	MYO1E			#614131	
181	Glomerulosclerosis, focal segmental, 5	null	INF2			#613237	
186	Glomerulosclerosis, focal segmental, 1	null	ACTN4			#603278	
201	Glomerulosclerosis, focal segmental, 2	null	TRPC6			#603965	
230	Other kidney disease causing nephrotic syndrome	null					
523	Membranous Nephropathy (MN)	null				#614692	
538	Minimal Change Disease (MCD)	null	PDCN; PT	ORPHA93	N04.0	#600995; #	
545	Focal Segmental Glomerulosclerosis (FSGS)	null	ACTN4;TR			#603278; #	

fill in the field "patientcount" with the no. of registered cases for each disease, save your file

Disease Matrix

Add Disease Scegli file Nessun file selezionato Save [Download Disease Matrix as XLS File](#)
[Download Disease Matrix as XLSX File](#)

Disease Name	Number of Patients, Donors	Gene	ORPHA Code	ICD 10	OMIM	Synonym(s)	Modified Date	Actions
Nephrotic syndrome, type 2	null	PDCN			#600995		06.03.2014 09:40:45	Actions
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Nephrotic syndrome, type 4	null	WT1			#256370		06.03.2014 09:43:47	Actions
Nephrotic syndrome, type 1	null	NPHS1	ORPHA839	N04	#256300		14.03.2014 14:06:00	Actions
Nephrotic syndrome, type 8	null	ARHGD1A			#615244		06.03.2014 09:58:18	Actions

upload your modified excel file (here, in Italian "Scegli file") and click "Save"

Standards

This area contains 8 fields to complete as appropriate:

- **Accreditation/certification program**
- **If yes, specify (ISO standards,...)**
- **Standardized case inclusion and exclusion criteria**
- **If yes, specify**
- **Standard Operating Procedures (SOPs) available for data management**
- **Quality control external audits**
- **If yes, frequency of audits**
- **Training program for the registering activities**

As your biobank/registry progresses through different stages you are able to edit this area as appropriate. The system will store **versions** of each set of Standards that you have saved over time, for you to reference in the future if required.

Accessibility

Complete the fields below as appropriate using a series of drop down and free text boxes:

- **Kind of Personal Data Collected**
- **Kind of data made available outside the registry**
- **Procedure to access data**
- **Other, please specify**
- **Has the registry a Data Access Committee?**
- **If yes, please provide the Data Access Committee webpage**
- **Do you use a Data Access Agreement?**
- **Data Access Agreement**
- **Are patient's data linked to other resources?**
- **Other, please specify**
- **Is informed consent mandatory as per your country's regulations?**
- **What kind of consent is obtained from patients/donors?**
- **Is an ethics board decision available for the use of data in research?**

As your biobank/registry progresses through different stages you are able to edit this area as appropriate. The system will store **versions** of each set of Standards that you have saved over time, for you to reference in the future if required.

Documents

Upload any appropriate documents (basic document, contract, marketing banner, online training, and sales presentations) to this area, to make available to anybody viewing your biobank/registry area on the Registry & Biobank Finder directory.

(Please note that if you do not add any information to the Standards, Accessibility or Documents area of the catalogue, these tabs will not appear on the live website. They only appear if an area contains information that has been entered).

Using the Registry & Biobank Finder directory

As well as adding your own biobank/registry to Registry & Biobank Finder, you are also able to view all other registries and biobanks that have been added. The catalogue homepage has five sections:

- Catalogue
- Recent Activities
- Propose
- Search
- Help Request

Logo	Name	Type	Number of Cases	Data Access	Request data	Number of access
	3q29 deletion Registry	Registry	38	not specified	jmulle@emory.edu	121
	Alagille Syndrome Alliance International Registry	Registry	136	not specified	alagille@alagille.org	61
	American Partnership for Eosinophilic Disorders	Registry	659	not specified	mjstobel@apfed.org	42
	Australian Myotonic Dystrophy Registry	Registry	0	not specified	hugh.dawkins@health.wa.gov.au	20
	Australian National Duchenne Muscular Dystrophy Registry	Registry	0	not specified	caroline.graham@health.wa.gov.au	31
	Australian National Spinal Muscular Atrophy Registry	Registry	0	not specified	caroline.graham@health.wa.gov.au	28

Catalogue

The catalogue tab lists all registries/biobanks that have been added by other members of RD-Connect, which you and external visitors to the catalogue can view. On the **General User Reporting System** page you can use the search box to enter information to find the correct biobank/registry that you would like to view. You can search by: the Disease Name, Gene, ORPHA Code, ICD10 or OMIM (information that was added to the Disease Matrix).

If your search term brings back a large number of search results, you can filter them by the type (biobank or registry) using the “Type” drop-down menu.

Recent Activities

The “Recent Activities” tab shows in chronological order, the registries and biobanks that have been recently edited within Registry & Biobank Finder.

The screenshot shows the 'Recent Activities' tab selected in the RD Connect interface. The page title is 'Linking up rare disease research across the world'. Below the navigation tabs (Catalogue, Recent Activities, Propose, Search, Help Request), there is a section titled 'Registries and Biobanks with recent activities'. Under this section, there are two categories: 'Updated Today' and 'Last update more than 2 months'. The 'Updated Today' category lists the 'GNE Myopathy Disease Monitoring Program (GNEM-DMP)' with contact information for Phillip Cammish and the website URL. The 'Last update more than 2 months' category lists the 'UK SMA Patient Registry' with contact information for Agata Robertson and the website URL, and the 'Global Fukutin Related Protein Registry' with contact information for the Coordinator and the website URL.

Propose

While logged in to Registry & Biobank Finder, you can create an additional registry or biobank using the **Propose** tab. This is the exact same process as used when registering for the first time— with the exception that the Submitter fields are automatically filled with your name and email address.

The screenshot shows the 'Propose' tab selected in the RD Connect interface. The page title is 'Linking up rare disease research across the world'. Below the navigation tabs (Catalogue, Recent Activities, Propose, Search, Help Request), there is a section titled 'Candidate Propose'. Under this section, there is a form titled 'Propose Candidate'. The form has several fields: 'Country' (dropdown menu with 'all' selected), 'Type' (dropdown menu with 'Biobank' selected), 'Acronym' (text input field), 'Name' (text input field with a red asterisk indicating it is required), 'Contact Person' (text input field with a red asterisk), 'Address' (text input field), 'Contact E-Mail' (text input field with a red asterisk), 'URL' (text input field), and 'Head of Registry/Biobank' (text input field).

Search

The “Search” tab allows you to search for biobanks/registries based upon the **search terms** that were entered in the Disease Synonym area, as shown below:

The screenshot shows the RDConnect interface with the search results for the term 'hibm'. The search returned 2 hits: Remudy and GNE Myopathy Disease Monitoring Program (GNEM-DMP).

Search

hibm

Search

Search for: hibm

Search returned 2 hits (Registries:2 /Biobanks:0)

Remudy Japanese patient registry for Neuromuscular diseases
En Kimura
<http://www.remudy.jp/>

Diseases GNE myopathy: Number of Patients, Donors 162
DiseasesSynonym: **hibm**/DMRV/Nonaka myopathy

gnemDMP GNE Myopathy Disease Monitoring Program (GNEM-DMP)
Phillip Cammish
<https://www.gnem-dmp.com/>

Diseases GNE Myopathy: Number of Patients, Donors
DiseasesSynonym: DMRV, Distal myopathy with rimmed vacuoles, GNE myopathy, **hibm2**, Hereditary inclusion body myopathy type 2, IBM2, Inclusion body myopathy type 2, Nonaka myopathy, Quadriceps-sparing myopathy

Help Request

Use the “Help Request” tab to contact the system administrator if you have questions about using Registry & Biobank Finder, or experience other problems.

The screenshot shows the RDConnect interface with the Help Request form. The form includes fields for email, request type, and subject.

Help Request

If you have any problems pleas let us know.

Your Email

Request

General Request

Subject

To contact the administrator directly, please email: rd-connect.wp2@iss.it