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The RD-Connect Registry & Biobank Finder: the online directory of existing rare disease registries and biobanks

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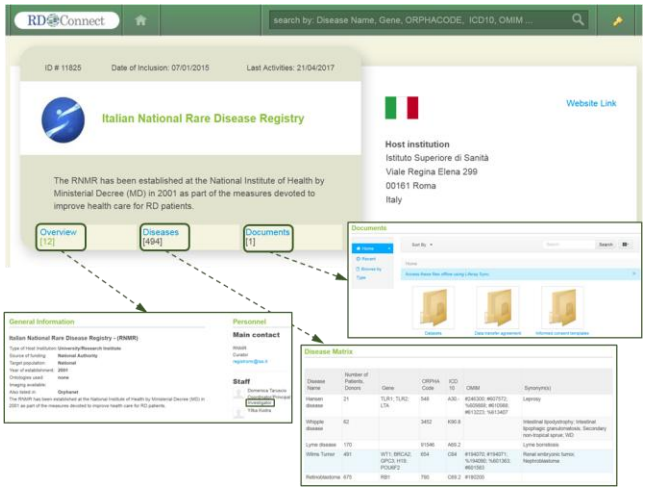
Introduction

Data on RD patients, including registry data and availability of bioresources are collected and stored in different biobanks and databases, and they are
usually not easily Findable, Accessible, Interoperable and Reusable (FAIR).
The **Registry & Biobank Finder** (http://catalogue.rd-connect.eu/) aims at concentrating sparse information on RD patients in one unique source by showing the number of samples/cases included in biobanks and registries, providing a first gateway towards more intense data sharing, and increasing the integration of biobanks and registries.
The RD-Connect Registry & Biobank Finder lets researchers find the rare disease patient registries and biobanks that store data on their rare disease of interest. The system provides databases' contact data and the numbers of registered cases for each disease in the registry/biobank, regularly updated by the database curators.
Aggregated data provided by the Finder, if appropriately checked, can be used by researchers who are trying to estimate the prevalence of a RD, to organize a clinical trial on a RD, or to estimate the volume of patients seen by different clinical centers.

Method

The information on a registry/biobank, is displayed in its individual **ID-Card**; each ID-Card consists in a webpage with information including contact information, aggregated data and metadata data such as accessibility and standards for the resources, standard operating procedures, documents for the informed consent, case report form. Importantly, the **"disease matrix"** (DM) section provides information on the number of patient cases and biological samples held for each RD, with its associated Orphacode, OMIM and ICD10 codes.

Figure 1. ID-Card of an individual registry



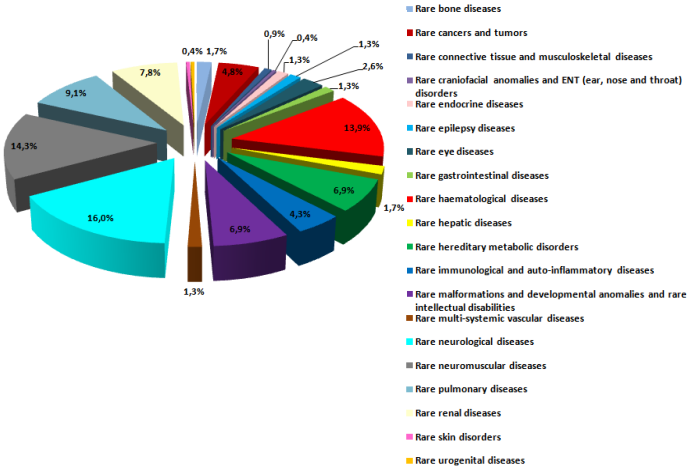
All users can browse the directory for the registries and/or biobanks for the RD of interest. Researchers can search the directory for biobanks or registries jointly or separately (filter by type) by biobank/registry name, disease name, ORPHAcode, OMIM and ICD-10 codes, synonyms, and other keywords related to the database.

Results

Currently Registry & Biobank Finder is populated with aggregated data for more than 230 registries and 22 biobanks of the EuroBioBank network.
Registries from all countries worldwide are participating, with a strong contribution from US (24,8%), followed by International registries (18%) Italian (12,6%), French (7,7%), German (5,9%), UK (5,4%) and Spanish registries (4,5%) and most of them belong to a specific network. National programs or platforms.

The most represented disease categories in RD registries are rare neurological diseases (16%), rare neuromuscular diseases (14%), rare hematological diseases (13,9%), rare pulmonary diseases (9,1%) rare renal diseases (7,8%) and rare hereditary metabolic disorders (6,9%)

Figure 2.



Conclusions

RD-Connect Registry & Biobank Finder enables more intensive sharing of patient disease information and biosamples. Database managers working on the same or related RDs can increase mutual visibility, paving the way to new scientific collaborations. It also allows researchers to find registries and biobanks storing data or biosamples for their disease of interest.

The classification of ID-Cards into disease areas based on the European Reference Networks (ERNs) has underlined the potential role of the Registry & Biobank Finder within the ERN community in order to increase the visibility of the registry activities within and to become an important tool according the European Union need in the registries field.

Gainotti S. et al.: The RD-Connect Registry & Biobank Finder: a tool for sharing aggregated data and metadata among rare disease researchers. 2018, Eur J Hum Genet.