Orphanet Report Series

Rare Diseases collection

April 2024

Rare Disease Registries, cohorts and databases

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Methodology and Objectives

Patient registries, cohorts and databases constitute key instruments to develop clinical research in the field of rare diseases (RD), to improve patient care and healthcare planning. They are the only way to pool data in order to achieve a sufficient sample size for epidemiological and/or clinical research, including natural history studies. They are vital to assess the feasibility of clinical trials, to facilitate the planning of appropriate clinical trials and to support the enrolment of patients.

Registries of patients treated with orphan drugs are particularly relevant as they allow the gathering of evidence on the effectiveness of the treatment and on its possible side effects, keeping in mind that marketing authorization is usually granted at a time when evidence is still limited although already somewhat convincing.

This report provides summarized data about rare disease registries, and it is based on the information collected by Orphanet so far, regarding systematic collections of data for a specific disease or a group of diseases, including patient registries, cohorts and databases.
The report includes data about countries participating to the Orphanet network. Registries, cohorts and databases funded by a member of IRDiRC (International Rare Diseases Research Consortium) are also taken into account, even if they are located in a country which is not part of the Orphanet network.

Data in this report reflects the last update made in the Orphanet database and may not depict changes occurred after this analysis.

In order to obtain the detailed list of registries, cohorts and databases collected by Orphanet with all the relative information, such as the disorder list covered and corresponding ORPHAcodes, the geographical location, geographical coverage and other descriptors, we invite you to visit Orphadata. This platform provides access to a number of massive datasets extracted from the Orphanet knowledge base, including datasets from our catalogue of expert ressources, upon signature of a Data Transfer Agreement or Service Contract.

Total number of registries, cohorts and databases and distribution by country

The number of registries, cohorts and databases collected by Orphanet up to April 2024 is 845. Figures 1 and 2 show the global distribution of these activities and the number of registries, cohorts and databases by country.

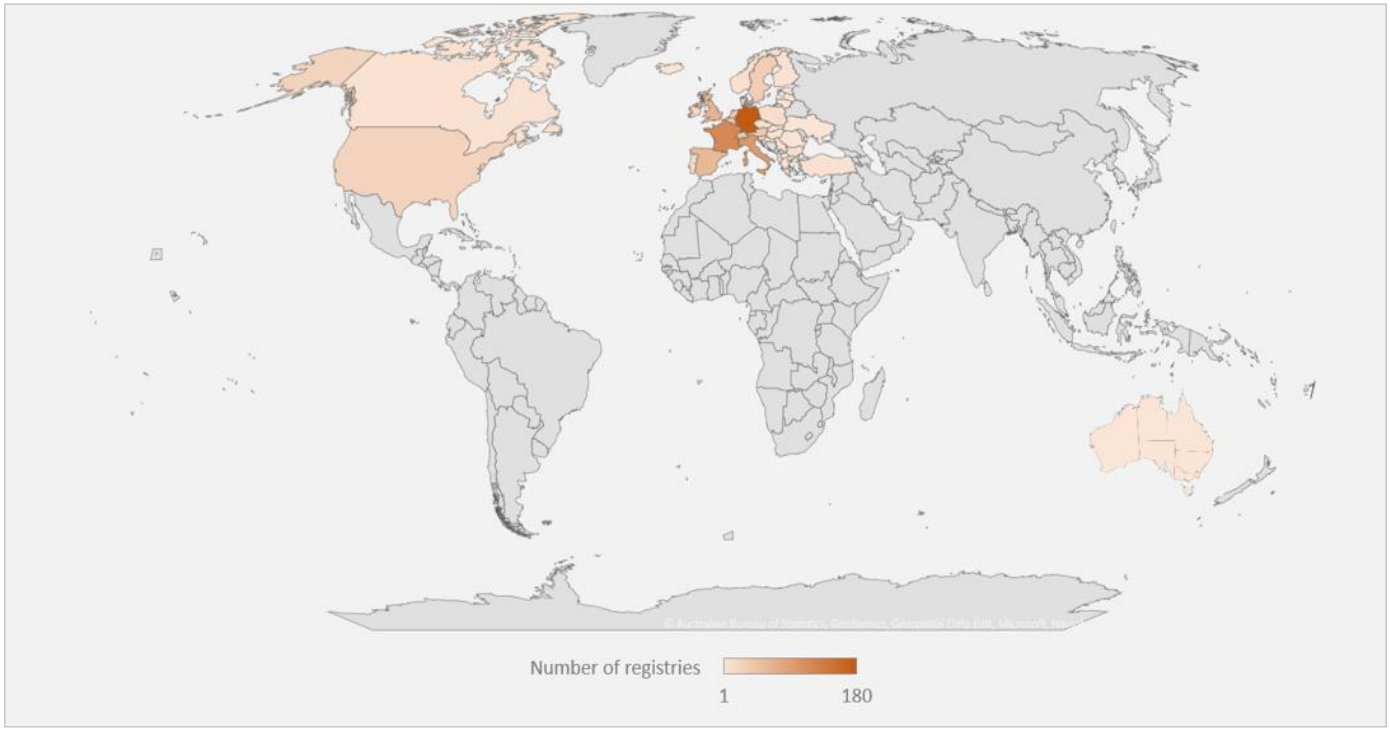

Figure 1. Global distribution of registries, cohorts and databases collected by Orphanet.

The country with the highest number of registries, cohort and databases is Germany (180), followed by France (120) and Italy (97).

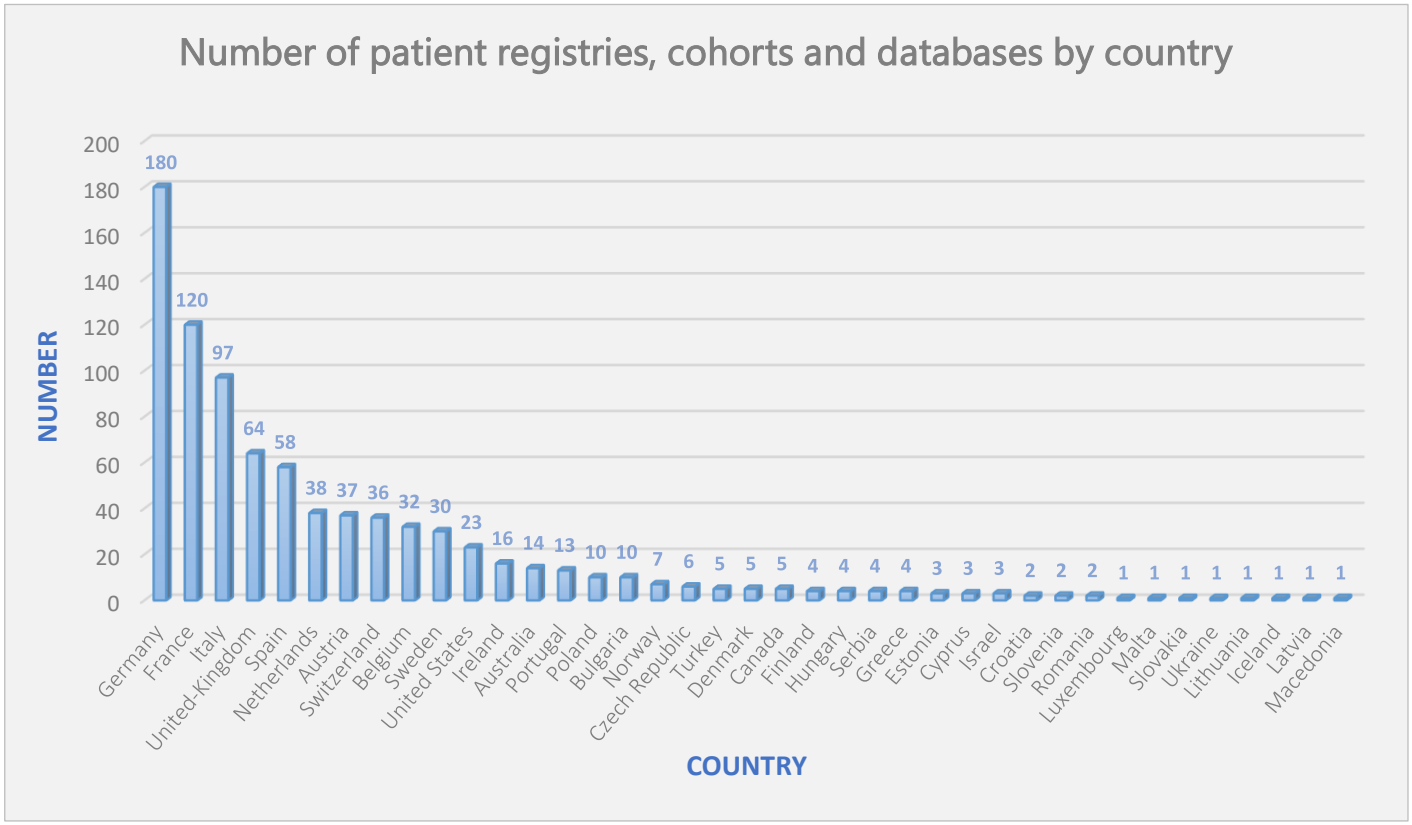

Figure 2. Number of registries, cohorts and databases collected by Orphanet by Country.

Active versus non-active patient registries, cohorts and databases

A patient registry, cohort or database is considered as non-active/terminated when:
- It is known to be terminated and the termination date could be retrieved.
- Its website has not been longer updated for longtime and/or their contacts cannot be contacted anymore.

Figure 3 shows that the vast majority of patient registries, cohorts and databases registered in Orphanet are still active.

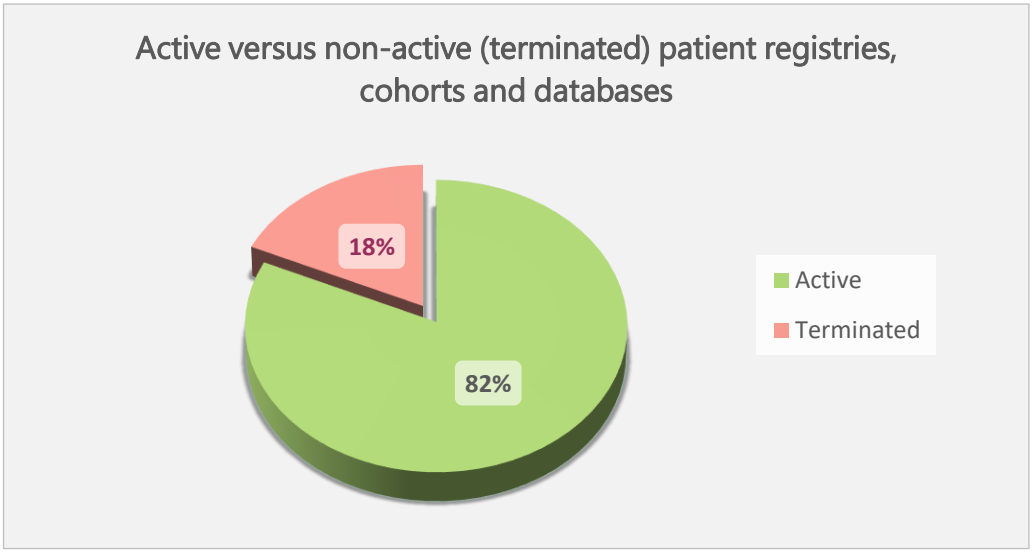

Figure 3. Distribution of active versus non-active patient registries, cohorts and databases.

Geographical coverage of patient registries, cohorts and databases

Figure 4 shows the geographical coverage of patient registries, cohorts and databases collected by Orphanet. The majority of these resources have a national coverage, while the proportion of patient registries, cohorts and databases with a regional, European or global coverage is very similar.

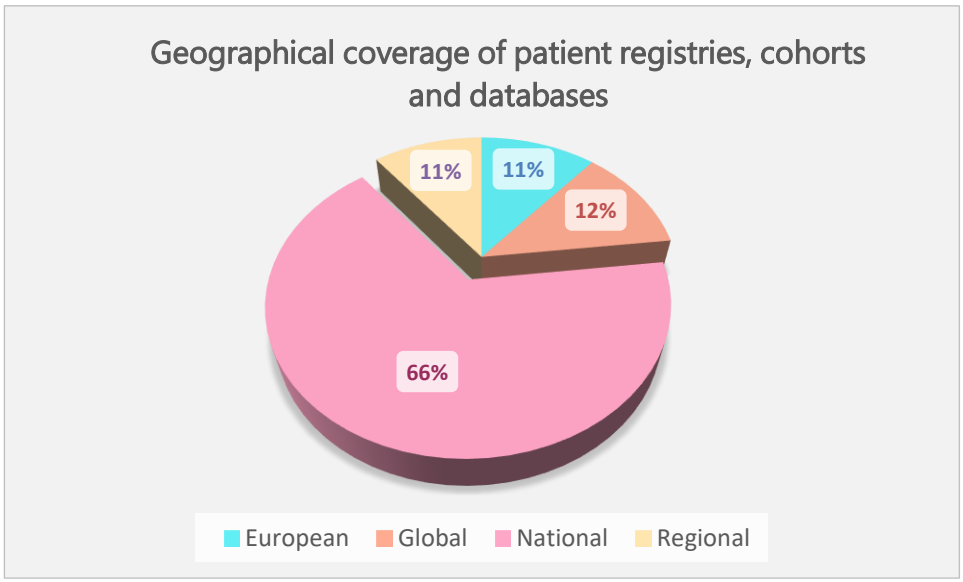

Figure 4. Geographical coverage of patient registries, cohorts and databases.

Coverage by medical domain

The total number of rare diseases or groups of rare diseases covered by patient registries, cohorts and databases is 1,061. Figure 5 shows the proportion of preferential medical domains covered by the patient registries, cohorts and databases collected by Orphanet. Of note, in the Orphanet classification, many diseases or groups of diseases can be assigned to more than one medical domain (due to the multidimensional nature of rare diseases) but only the preferred domain (determined according to published procedures) is presented in this figure.

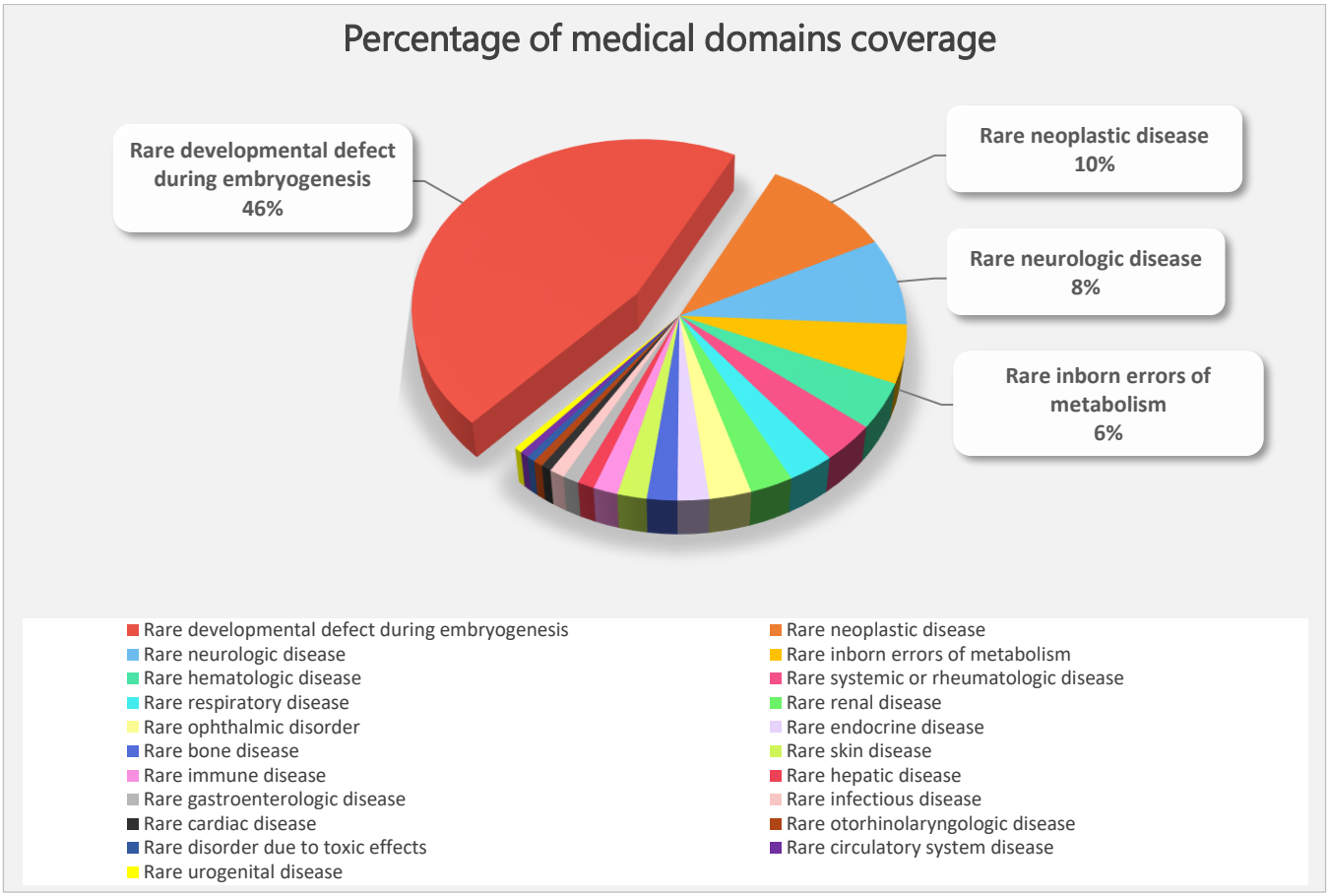

Figure 5. Proportion of preferential medical domains covered by patient registries, cohorts and databases. The percentage above 5% are indicated in the figure. Medical domains shown in the figure are in decreasing order.

As shown in the figure, the medical domains most represented (above 5%) are rare development defect during embryogenesis (46 %), rare neoplastic disease (10 %), rare neurologic disease (8 %) and rare inborn errors of metabolism (6%).

Rare diseases or groups of rare diseases with the highest number of patient registries, cohorts and databases

The analysis of the rare diseases or groups of rare diseases covered by patient registries, cohorts and databases shows that cystic fibrosis, with 36 registries, cohorts or databases, is the most represented rare disease.

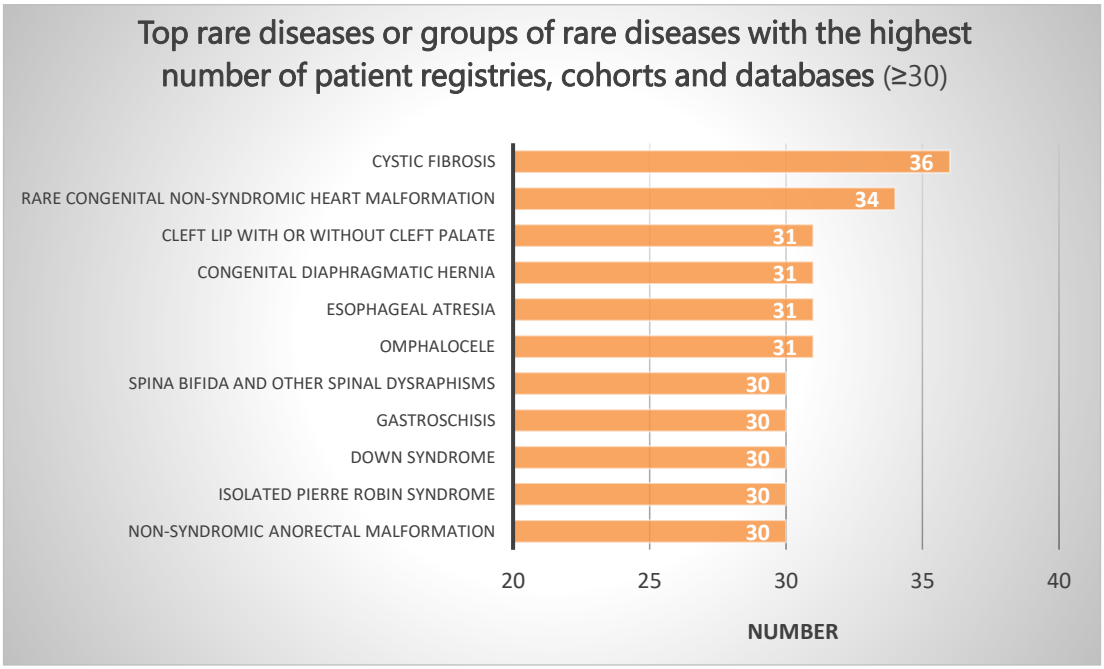

Figure 6. Top rare diseases or groups of rare diseases with the highest number of patient registries, cohorts and databases.

Number of patient registries, cohorts and databases funded by an IRDiRC member

Registries, cohorts and databases funded by a member of IRDiRC are collected by Orphanet, even if they are located in a country which is not part of the Orphanet network. To know the list of IRDiRC members, we invite you to visit the IRDiRC website. As shown in figure 6, 18% of patient registries, cohorts and databases are funded by an IRDiRC member.

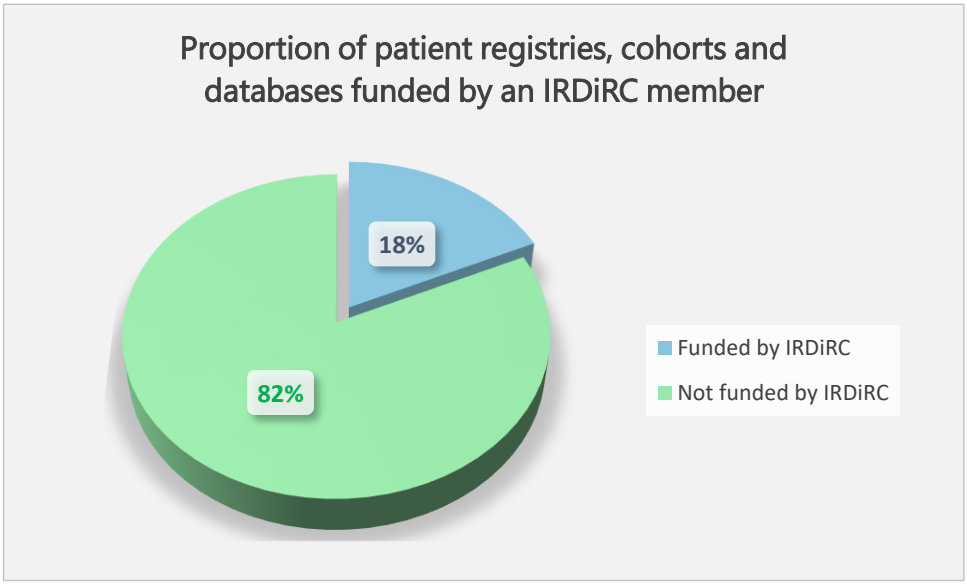

Figure 7. Proportion of patient registries, cohorts and databases funded by an IRDiRC member.

Proportion of patient registries, cohorts and databases by funding body types

Among patient registries, cohorts and databases with a specified funding body, Figure 8 shows that 56% are funded by a public funding body and 44% by a private funding body (10% for-profit and 34% non-for-profit).

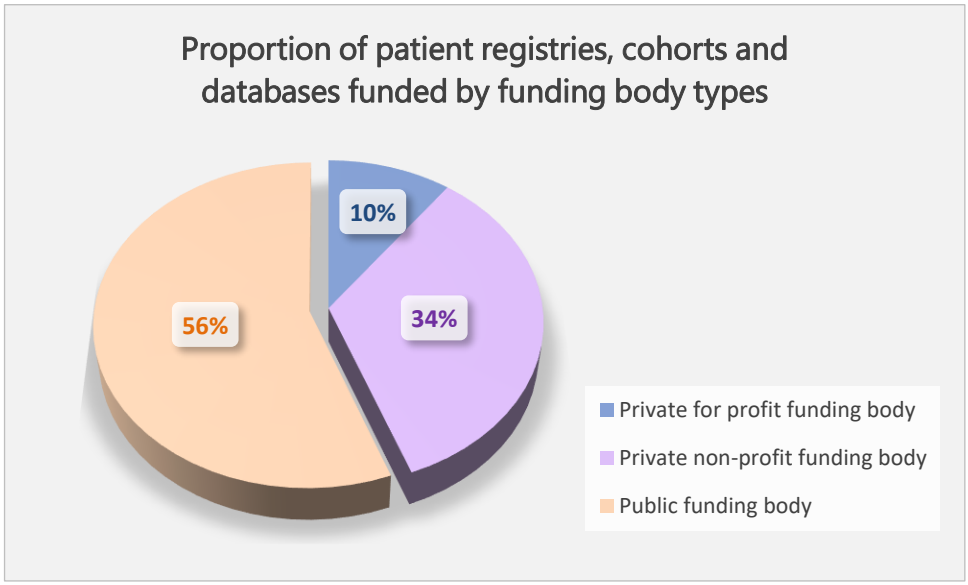

Figure 8. Proportion of patient registries, cohorts and databases by funding body types.

ERN registries

In order to support the development of registries and databases in the field of rare diseases for epidemiological purposes and to promote the interoperability of data in rare diseases registries, the European Commission co-funds the 24 European Reference Networks (ERNs) to develop their research capabilities, including the construction of patient registries.

The development of these European patient registries aims to gather sufficient patient data to follow up the natural course of diseases.

In the table 1, the list of the 24 ERNs registries with the relative coordinating country is shown.

ERN patient registry name	Coordinating country
ERN [BOND] - EuRR-Bone: European Registry for Rare Bone and Mineral Conditions	Netherlands
ERN [ERKNet] - ERKReg: The European Rare Kidney Disease Registry	Germany
ERN [CRANIO] - ERN CRANIO registry	Netherlands
ERN [EpiCARE] - EPICARE-GRANT: An Operational EPICARE Registry for all Rare and Complex Epilepsies	Belgium
ERN [ERN-LUNG] - REGISTRY WAREHOUSE - RD Registry Data Warehouse	Germany
ERN [EuroBloodNet] - ENROL: European Rare Blood Disorders Platform	Spain
ERN [EURACAN] - STARTER: STarting an Adult Rare Tumor European Registry	Italy
ERN [ERNICA] - ERNICA-registry for improving care	Netherlands
ERN [GENTURIS] - GENTURIS registry: The Genetic Tumour Risk Syndromes Registry	Netherlands
ERN [eUROGEN] - ERN eUROGEN Registry for rare urogenital diseases	Netherlands
ERN [EYE] - REDgistry: An interoperable sustainable European Rare Eye Disease Registry	France
ERN [EURO-NMD] - EURO-NMD Registry: Patient centered and interoperable registry hub for Rare Neuromuscular Diseases	France
ERN [PAEDCAN] - PARTNER: Paediatric Rare Tumours Networks - European Registry	Italy
ERN [GUARD-Heart] - The Heart-Core Registry: a Gateway to Uncommon and Rare Diseases of the Heart	Netherlands
ERN [ITHACA] - ILIAD Rare Diseases patient registry: an International Library of Intellectual disability and Anomalies of Development	France
ERN [RARE-LIVER] - R-LIVER: Registry for Rare Liver Diseases of the ERN on hepatological diseases	Germany

ERN [TRANSPLANT-CHILD] - PETER: PaEdiatric Transplantation European Registry	Spain
ERN [ReCONNET] - TogethERN ReCONNET: a European Registry Infrastructure for data harmonization in rare and complex connective tissue and musculoskeletal diseases	Italy
ERN [RITA] - MERITA: A METADATA REGISTRY FOR THE ERN RITA	Italy
ERN [RND] - ERN-RND Registry: The ERN-RND Rare Neurological Disease Registry	Germany
ERN [VASCERN] - VASCERN Registries	France
ERN [Skin] - ERN-Skin REGISTRY: Interoperable ERN on Rare and Undiagnosed Skin Disorders	France
ERN [MetabERN] - U-IMD: Unified European Registry for Inherited Metabolic Disorders	Germany
ERN [Endo-ERN] - EuRRECa: European Registries for Rare Endocrine Conditions	United Kingdom

Table 1: List of the 24 ERNs registries

Please note that all data presented in this report are available for download at www.orphadata.com

For any questions or comments, please contact us: contact.orphanet@inserm.fr

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The correct form when quoting this document is:

« Rare Disease Registries, cohorts and databases », Orphanet Report Series, Rare Diseases collection, April 2024

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