# National RD registry in Spain: Pilot study of the Spanish Rare Diseases Registries Research Network (SpainRDR)

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## AIMS

The development of a national Rare Diseases (RD) registry in Spain was launched in 2012 with the project SpainRDR, supported by the International Rare Diseases Research Consortium (IRDiRC)

SpainRDR includes two different strategies: patient registries addressed to patient outcome research and population-based registries addressed to epidemiologic research, health and social planning

The pilot study aims to detect the difficulties of developing the national and population-based RD registry

## METHODS

Both comprehensive RD lists and common data elements (CDE) have been defined and harmonized with other international strategies (EPIRARE, RD-CONNECT, NIH). CDEs mainly comprise variables related to personal identification data and RD definition

RD patient information was collected from regional health databases corresponding to 2010 and 2011: electronic hospital records (discharges basic minimum dataset), mortality registry, health insurance card databases, electronic primary care clinical records, chronic renal diseases registry, orphan drugs registry, newborn screening registry and tumor registry, among others

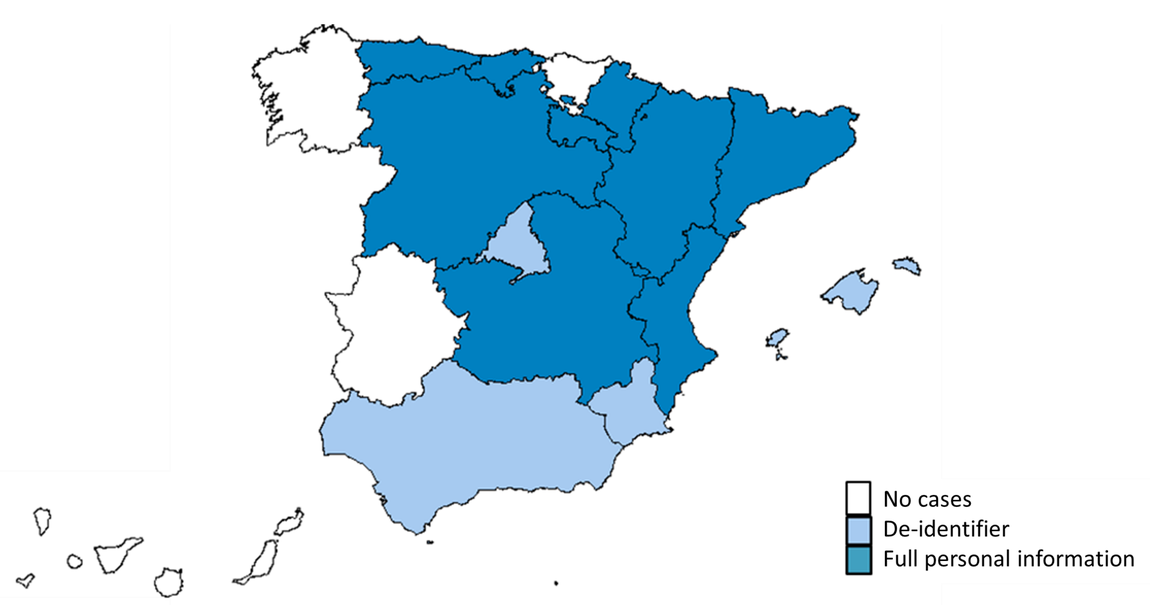


**Figure 1.** Participation of Regional Health Departments in the pilot study: types of reported cases to central repository

## RESULTS

Data representing 80.2% of the Spanish population have been initially communicated to the central data repository during the pilot study. A total of 824,399 RD cases have been detected

As an example, RD show 26% congenital anomalies; 19% endocrine, nutritional and metabolic diseases; 13% blood and blood-forming organs; 10% diseases of the circulatory system

Practical problems detected in the pilot study have been discussed and fixed. Final patient recruitment has already started and it will include RD cases detected from 2010 to 2012

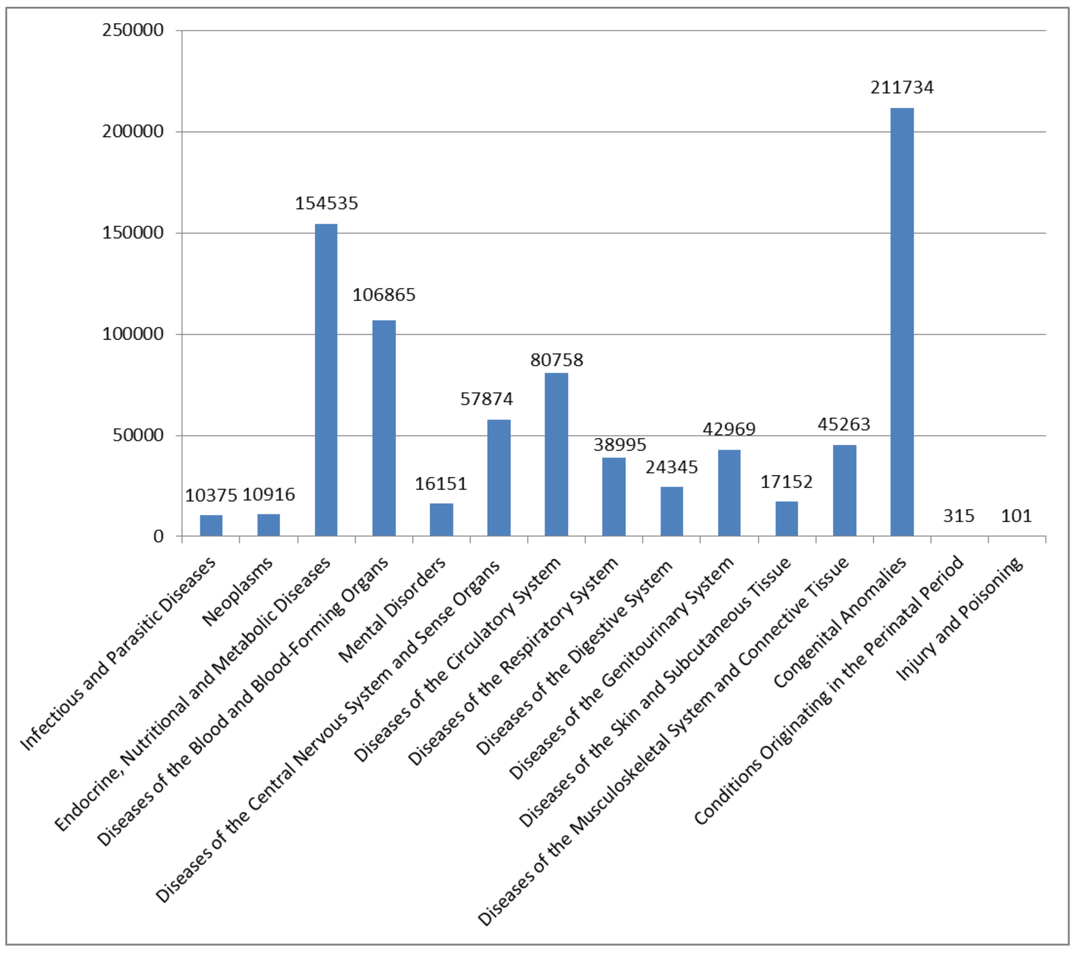


**Figure 2.** Number of RD cases by ICD chapter

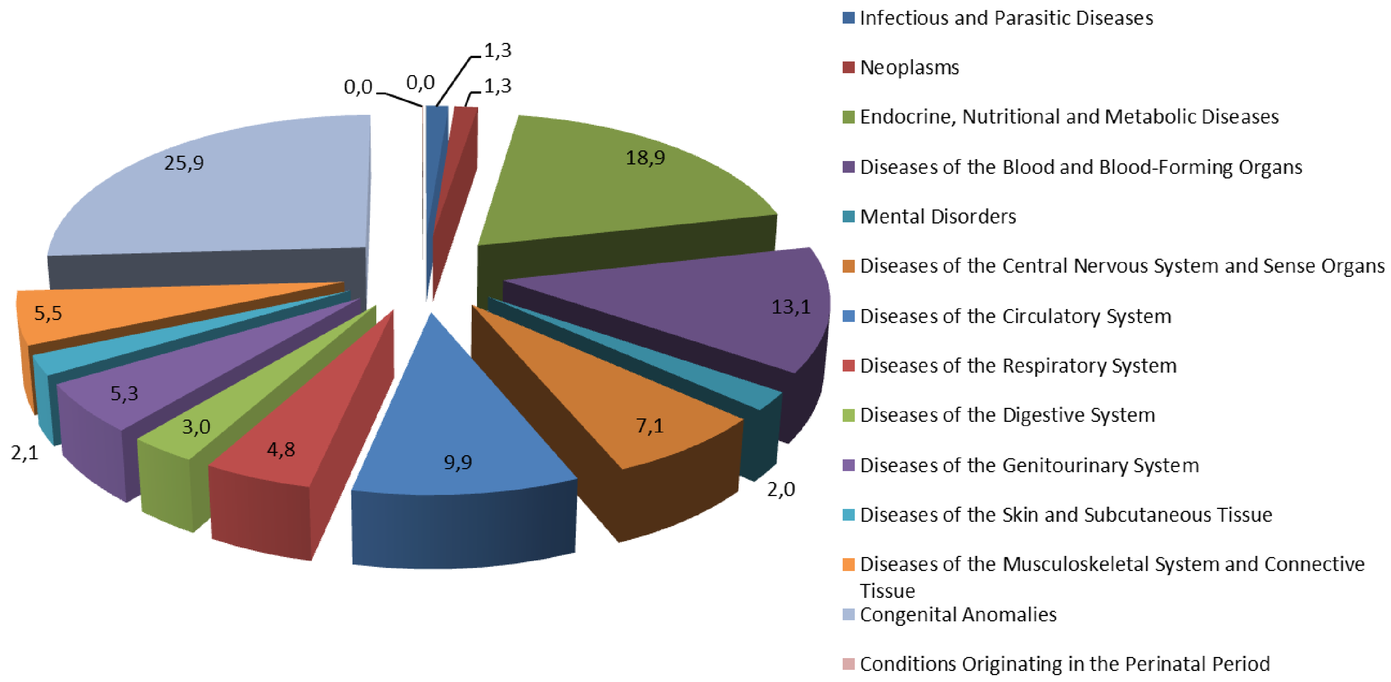


**Figure 3.** Percentage of RD cases by ICD chapter

**Table 1.** Example of RD detected and number of cases

| RD | Cases | RD | Cases |
|---|---|---|---|
| ICHTHYOSIS CONGENITA | 1114 | WEGENER'S GRANULOMATOSIS | 1004 |
| OSTEOGENESIS IMPERFECTA | 575 | POLYARTERITIS NODOSA | 3505 |
| MARFAN'S SYNDROME | 707 | SJOGREN'S DISEASE | 7550 |
| AMYOTROPHIC LATERAL SCLEROSIS | 2209 | GIGANTISM AND ACROMEGALY | 887 |
| HUNTINGTON'S DISEASE | 782 | MYASTHENIA GRAVIS | 387 |
| POLYCYTHEMIA VERA | 3381 | IDIOPATHIC PULMONARY FIBROSIS | 2891 |
| MYCOSIS FUNGOIDES | 2783 | EHLERS-DANLOS SYNDROME | 189 |
| BEHÇET'S DISEASE | 1167 | GENERALIZED SCLERODERMA | 3587 |

## CONCLUSIONS

In summary, National Institute of Rare Diseases Research and Regional Health Departments of Spain are working together towards a harmonized RD patient registration

The Spanish experience could be a model for other countries with complex political and administrative structures which, in order to carry out a national RD registry, will require the standardization of criteria, data harmonization and coordination between regions

SpainRDR Spanish Rare Diseases Registries Research Network