

POSTER PRESENTATION

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National rare diseases registry in Spain: pilot study of the Spanish Rare Diseases Registries Research Network (SpainRDR)

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Background

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

Material and 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.

Results

Data representing 80.2% of the Spanish population have been initially communicated to the central data repository during the pilot study (Figure 1). 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 and certain disorders involving the immune mechanism; 10% diseases of the circulatory system (Figure 2). 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.

Conclusion

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.

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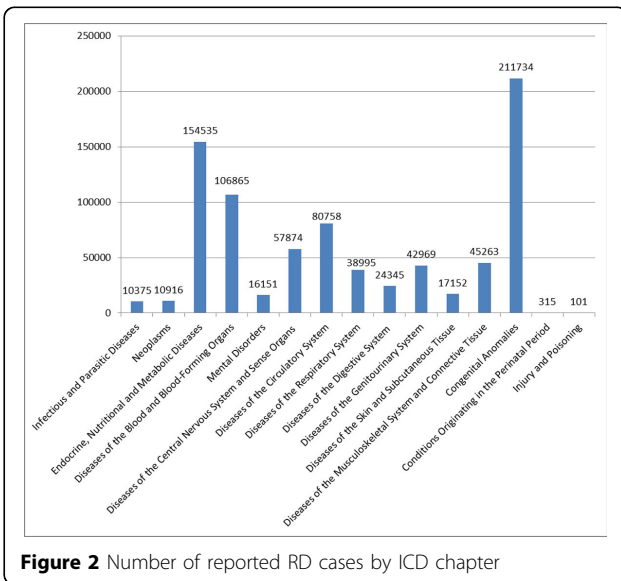
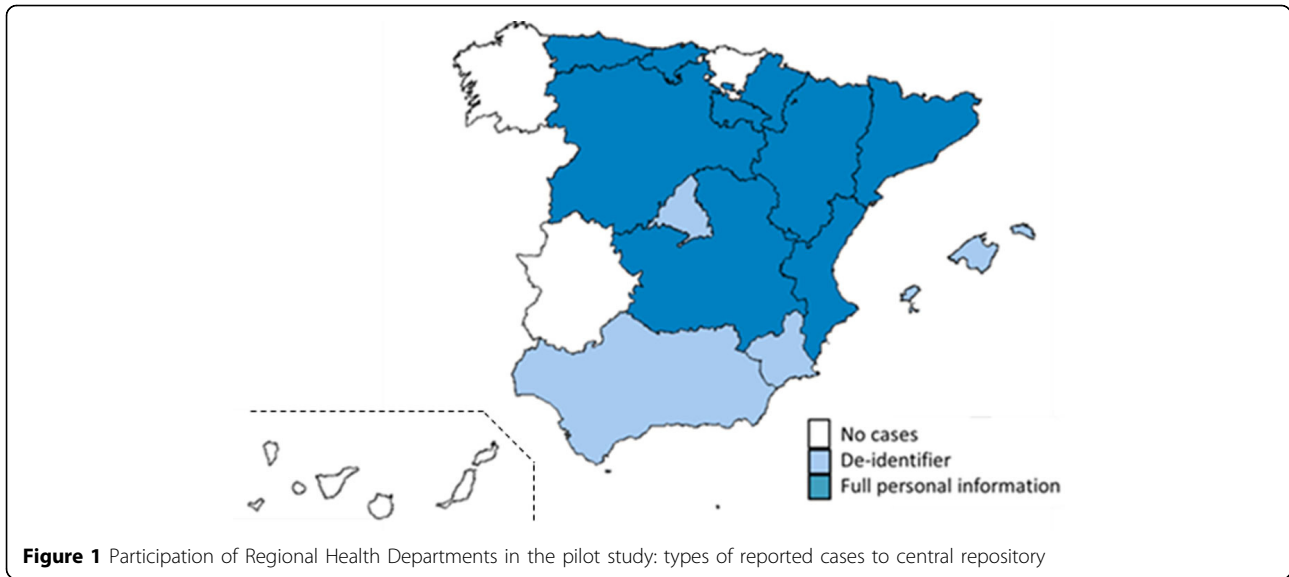
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