Introduction

In 2009 the European Commission (EC) released recommendations on actions in the field of rare diseases1. These were to:
1. Establish and implement national plans or strategies for rare diseases
2. Define, code and inventory rare diseases
3. Generate and contribute to research on rare diseases
4. Create rare disease centres of expertise
5. Gather national expertise on rare diseases
6. Empower patient organisations
7. Ensure sustainability of infrastructure for rare diseases

The objective of this analysis was to review the policies in place for rare diseases before the EC recommendations, and evaluate levels of achievement of the policies in meeting the EC recommendations, in particular, focusing on EU collaborative research, neonatal screening and the number of rare disease registries per country.

Methods

National plans on rare diseases created by EU countries2 (and available in the English language) were identified. The policy context pre and post the 2009 EC recommendation was assessed for these countries based on the 2009 European Union Committee of Experts on Rare Diseases (EUCERD) Report3. Subsequent EC reports in 20114 and 20165 were also used to establish levels of achievement in implementing the 2009 EC recommendation.

Results

National plans were available for 16 countries2. Prior to the 2009 EC recommendation, there were broad differences in policy regarding rare diseases throughout the EU (Table 1). Following the 2009 EC recommendation5, all countries included in our analysis created a national plan or strategy which closely followed the recommendation (data not shown)6.

EU Collaborative Research

Seven countries did not report collaborating on rare disease research prior to the 2009 EC recommendation, while all 16 countries reported collaborating after the EC recommendation2,3. Of the 16 countries, Germany took part in the most studies (115), followed by France, Italy and the UK (103, 95 and 84 studies, respectively); while Bulgaria took part in the fewest (2 studies) (Figure 1).

Neonatal Screening for Rare Diseases

Prior to the EC recommendation, all 16 countries had neonatal screening programmes in place (data not shown)3. A subsequent report in 2011 showed that of the 16 countries in our study, the leading country for rare disease screening was Austria, with a total of 29 rare diseases regularly screened for at birth, followed by Spain (27 diseases) and Hungary (25 diseases) (Figure 2)6. The majority of the rare diseases screened were metabolic disorders (76%)7.

Rare Disease Registries

Before the EC recommendation, six countries had no rare disease registries3. However, a 2016 report showed that all countries have subsequently created national level registries for rare diseases (Figure 3)8.

Conclusions

Our research has highlighted that the degree of adherence to the EC recommendations is varied, and that irregularity in the rare disease landscape throughout the EU remains an issue. Despite this, the EC recommendation provided a structure for countries when developing their rare disease strategies, and has proven effective through the implementation of national plans. The needs of patients with rare diseases are now stated as a priority for EU countries, with the majority of the seven recommendations being achieved in some form. Although the EC recommendation highlighted the need for uniformity in a number of fields, the recommendation fails to directly address policies on the reimbursement of treatments for rare diseases which would ensure timely, equitable and sustainable access to medicines for patients with rare diseases across the EU.

A limitation of this study is that not all national plans were available. Further research could seek to understand the cause of variation in rare disease strategies across the EU by exploring data such as gross domestic product, health care expenditure per capita, population size and the prevalence of rare diseases in each country.