Consensus on the criteria needed for creating a rare-disease patient registry. A Delphi study

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ABSTRACT

Background Patient registries (PRs) are important tools for public-health surveillance and rare-disease research. The purpose of this study is to identify the most important criteria for the creation of a rare-disease PR that could be used by public-health authorities to develop health policies.

Methods A consensus-development Delphi study was used, with participants selected for their expertise in rare diseases and registries. Participants were asked to complete a questionnaire on the most important criteria for creating PRs. Three rounds were performed.

Results Agreement was reached on half the questions in the first round and on 89% of questions in the final round, with a total expert participation rate of around 60% by the final stage. This study made it possible to reach a broader consensus starting from experts’ initial assessment of the features that should be considered for the creation of a rare-disease PR.

Conclusion The consensus method used made it possible to define the characteristics of a PR based on expert opinion within a rare-disease framework. This study may serve as a guide for helping other researchers plan and build a rare-disease PR.

Keywords Delphi technique, expert opinion, rare diseases, registry

Introduction

Rare diseases (RDs) are defined by the European Union (EU) as life-threatening or chronically debilitating diseases with low prevalence (<5/10,000).¹ There are estimated to be between 5000 and 8000 different RDs.² Their low prevalence creates difficulties and problems for RD patients and their families because there is little knowledge in this field and the public and private sectors invest insufficient resources.

A registry is a tool for improving knowledge on RDs³ and consists of a systematic compilation of data for the purpose of healthcare planning, implementation and evaluation in a defined population. Registries differ from one another depending on their target entities, the populations covered, the type of data collected and their main uses.⁴ The emergence of chronic diseases as a public-health concern initiated an increase in patient registries (PRs) in the 1950s that continues to
PRs have been used in recent years for the study of some RDs, such as cystic fibrosis, ne... PRs have been used in recent years for the study of some RDs, such as cystic fibrosis, neurofibromatosis, spina bifida, juvenile neuronal ceroid lipofuscinosis and ophthalmic dysphoria, among others. However, stakeholders in RDs (clinical professional societies, healthcare organizations, private or public research foundations, patients’ associations, their families and services) have not used existing systems to create a suitable RD registry. This could be due to the usual confusion between the concepts of ‘PR’, ‘population-based registry’ (PBR) and ‘disease registry’, although, according to the scientific literature, there are well-established differences. A PBR is characterized by defined populations, whereas a PR addresses specific diseases and does not have completeness as its main objective.

The ideal design and scope of a registry is determined by its intended purpose. It is important to identify key stakeholders and assess feasibility. Once the decision to proceed has been taken, the next essential issue is setting up a registry team, establishing its governance and oversight plan, defining the scope and rigor needed; and defining the dataset, patient outcomes and target population. Two further basic issues should be considered: the completeness of case ascertainment, and the validity of values for each data point. First, to minimize these limitations, best-practice methods and processes must be employed. Secondly, multiple data sources are preferable as they help experts to fully understand the natural history of the disease, and guarantee high-quality data.

The purpose of this study is to identify RD experts’ points of view on the most important characteristics and criteria for the creation of a PR that could be used by public-health authorities to develop health and social policies.

**Methods**

The aim of this study was to reach a consensus regarding the items necessary for creating a PR. To this end, a qualitative technique—a modified Delphi method—was used to conduct a national survey among RD experts to find out which patient-specific outcomes and features they considered essential for this kind of registry. The Delphi method is an iterative facilitator-led process that can take place without panellists needing to be physically present. Thus, its anonymous nature avoids the problem of ‘single influence’ in which a study’s results are influenced by one individual’s replies. The process went through the following stages.

**Selecting the panellists**

Panellists were recruited either for their experience or their prior participation in RD-related activities. The sample group was composed of 41 multidisciplinary experts. Most of them (18) were representatives of the Spanish Rare Disease Registry Research Network (SpainRDR), which is conducting a national project to create a Spanish RD registry. One expert was from the Centre for Biomedical Network Research on Rare Diseases (CIBERER)—a Spanish network structure promoting clinical excellence in RD; two were experts from the Spanish Rare Disease Federation (FEDER), which brings together more than 200 RD patients’ and family associations; five experts work in the Spanish Health Ministry; another five were selected from other scientific associations; four more were experts on health information systems; and the remaining six were experts on disease registries and information systems.

**Designing the Delphi items and survey**

The questionnaire’s objectives, scope and methodology were defined by the study’s steering committee, which was composed of the SpainRDR research team. The committee pondered and selected items it considered to be either the most important ones for creating a PR, or those that most commonly cause confusion. These items were previously identified through a literature search in specialist journals, policy documents and websites. A modified Delphi was then used that was based on a structured questionnaire that had 18 questions: 7 single-choice, 10 multiple-choice and 1 open question (Appendix 1). Space was provided next to each question to allow participants to add comments.

**Preparing and sending the questionnaires to the experts**

The study was conducted in three rounds. The questionnaire was built with ‘Google Docs’ (Google Inc.) and delivered via e-mail. Responses were collected during 30 days, with a reminder being sent 15 days after the first mailing. Responses were anonymized in each round.

In the first round, experts were requested to provide their opinions on the items presented to them. Some questions were found to be controversial and yielded variable responses, so they were reformulated in the second round to clarify their meaning. The multiple-choice questions asked participants to rate each item from the list on a 10-point scale (from 1—least important to 10—most important), and the opinion questions used a 5-point level-of-agreement Likert scale (from 1—strongly disagree to 5—strongly agree). The third round informed respondents of each item’s score in the previous
round and engaged them in a general discussion on those questions regarded as controversial, with the objective of reaching a final consensus.

**Analysing the Delphi data**

After each round a report was written to evaluate the round’s results and reformulate questions if necessary. Answers were considered appropriate when chosen by more than 50% of respondents. Answers with a lower percentage were deemed controversial and were included in the following round, to be assessed again by respondents.

In the case of the single- and multiple-choice questions, we obtained absolute frequencies, percentages and their 95% Bayesian credible intervals (95% CI) for the posterior probability of choosing each answer. The Bayesian credible intervals were based on the binomial model for the proportion, with $\beta (0.5, 0.5)$ prior distribution, and they were chosen from the commonly used confidence intervals due to their better accuracy. In the case of the rating-scale questions, we obtained quartiles, sample mean and standard deviation, and 95% Bayesian credible intervals for the mean. The 95% CIs were based on the normal model, with prior joint distribution of the mean and variance proportional to the inverse of the variance.

**Results**

The first round of the study was conducted in November 2012, with 25 (61%) of the experts surveyed completing the round. The second round took place in January 2013, with 59% of experts participating (24), and the third and final round was performed in February 2013 with the collaboration of 28 experts (68%). Consensus was reached on 50% of responses in the first round and finally on 16 of the 18 questions assessed, as shown in Table 1. Tables 2–4 show the results of the single, multiple-choice and rating-scale questions.

Question 1 of the first round was designed to test experts’ level of knowledge regarding the definition of PR. Responses show that 76% of experts agreed that 'PR' and 'PBR' have different meanings, and 68% said the same about ‘disease registry’ and ‘PBR’. The highest discrepancy was observed when defining ‘PR’ and ‘disease registry’: there was no consensus for these terms, with 56% of respondents answering that they were almost synonymous but with just a slight difference. A reformulated version of this question was, therefore, carried over into the second round. It assessed the level of agreement on five definitions of PR, with 88% of experts choosing the correct answer (see Table 4), a result that was confirmed in the third round.

Table 3 was an open question meant to collect different opinions that were then turned into a single-choice question

<table>
<thead>
<tr>
<th>Q</th>
<th>Item</th>
<th>First round</th>
<th>Second round</th>
<th>Third round</th>
<th>Final consensus</th>
</tr>
</thead>
<tbody>
<tr>
<td>1</td>
<td>Definition of PR</td>
<td>X</td>
<td>X</td>
<td></td>
<td>X</td>
</tr>
<tr>
<td>2</td>
<td>Criteria for building and maintaining a registry</td>
<td>X</td>
<td></td>
<td></td>
<td>X</td>
</tr>
<tr>
<td>3</td>
<td>Most important characteristic of a disease for it to be included in an RD registry</td>
<td>X</td>
<td>–</td>
<td>–</td>
<td></td>
</tr>
<tr>
<td>4</td>
<td>Fundamental purpose of a registry</td>
<td>X</td>
<td></td>
<td></td>
<td>X</td>
</tr>
<tr>
<td>5</td>
<td>Type of population that should be included in a registry</td>
<td>X</td>
<td>X</td>
<td></td>
<td>X</td>
</tr>
<tr>
<td>6</td>
<td>Preferred geographic area</td>
<td>X</td>
<td></td>
<td></td>
<td>X</td>
</tr>
<tr>
<td>7</td>
<td>RDs that are most needed in an PR</td>
<td>X</td>
<td>X</td>
<td></td>
<td>X</td>
</tr>
<tr>
<td>8</td>
<td>Key features</td>
<td>X</td>
<td></td>
<td></td>
<td>X</td>
</tr>
<tr>
<td>9</td>
<td>Information that must be included in a registry</td>
<td>X</td>
<td>X</td>
<td></td>
<td>X</td>
</tr>
<tr>
<td>10</td>
<td>Outcomes that should be included</td>
<td>X</td>
<td></td>
<td></td>
<td>X</td>
</tr>
<tr>
<td>11</td>
<td>Intermediate outcomes</td>
<td>X</td>
<td></td>
<td></td>
<td>X</td>
</tr>
<tr>
<td>12</td>
<td>Final outcomes</td>
<td>X</td>
<td></td>
<td></td>
<td>X</td>
</tr>
<tr>
<td>13</td>
<td>Advantages of an PR</td>
<td>X</td>
<td>X</td>
<td></td>
<td>X</td>
</tr>
<tr>
<td>14</td>
<td>Minimum staff that a registry should have</td>
<td>X</td>
<td></td>
<td></td>
<td>X</td>
</tr>
<tr>
<td>15</td>
<td>Most appropriate institution for a registry</td>
<td>X</td>
<td></td>
<td></td>
<td>X</td>
</tr>
<tr>
<td>16</td>
<td>Entities that provide access to outcomes</td>
<td>X</td>
<td></td>
<td></td>
<td>X</td>
</tr>
<tr>
<td>17</td>
<td>Form in which data should be transferred</td>
<td>X</td>
<td>X</td>
<td></td>
<td>X</td>
</tr>
<tr>
<td>18</td>
<td>Registry information that should be made public</td>
<td>X</td>
<td>–</td>
<td>–</td>
<td>–</td>
</tr>
</tbody>
</table>

Q, question; X, yes; –, no.
in the second round, with the following items: disease prevalence, availability or not of treatment, disease specificity, patient age, disability and quality of life, and genetic component. Surprisingly, the option ‘disability and quality of life’ obtained consensus in this round, but not in the third round.

Regarding the type of population to be included in the registry (question 5), experts agreed (68%) on the use of both types (‘reference population’ and ‘managed reference population’). Concerning the types of RD that should be included in a registry (question 7), no consensus was reached in the first round, so the question was completely reformulated in the three sections of the second round. The following characteristics obtained a consensus in the second round: prevalence above 0.1/10 000 (50%), the availability of a specific orphan drug (67%) and the disease being one of the endocrine, metabolic or nervous system (79%). This question was eventually accepted in the third round.

The items selected by experts as the most important ones for creating a registry (question 9) were date of birth (92%) and sex (92%). Other items also obtained 50–75% scores, however, so in order to reach a broader consensus this question was reformulated into a single-choice question in the next round. The items that came out of the second round as most important were personal health identification (ID) number (33%) and date of birth (25%). These results were confirmed in the third round.

When asked about the benefits offered by a registry (question 13), 50% of experts marked all items as potential benefits, with very similar scores for all of them. This question was, therefore, reformulated in the second round, with each potential benefit being assessed on a 1–5 rating scale. The item that was eventually considered to be most important advantage of a registry was ‘Serves as a tool to locate or identify cases for researchers and patients interested in participating in studies or in clinical or therapeutic trials’.

Question 15 inquired about the most appropriate environment for locating a PR, with the option ‘Health authority’ receiving most of the votes (64%). There was no consensus, however, regarding whether this should be a regional (32%) or a national entity (32%).

The highest discrepancy in results was found in question 17, which dealt with how results should be transferred. No consensus was reached on the type of anonymization that should be used. This question was, therefore, included in the second round, where ‘data anonymization with coded identification’ obtained a 50% score.

Finally, when presented with a list of types of information that a registry should make public (question 18), experts decided in favour of all of them. We, therefore, asked them to rate the items again in the second round and this time ‘Lethality’ was considered the most important one, with a total score of 160 points. In the third round, however, 79% of experts failed to confirm this, so this question ended up being one for which no consensus was obtained.

**Discussion**

**Main study findings**

The study showed there was broad agreement among experts on the characteristics that should be considered in the creation of a rare-disease PR, and made it possible to extract different conclusions from their opinions. The consensus method...
employed enabled experts from different specific fields to reach agreement on the definition of a PR in the context of RDs, as well as on the criteria that a RD should meet in order to be included in such a PR. This study may, therefore, help other researchers to plan, create and manage a PR of RD.

Current knowledge on the subject

Currently, there are so many different concepts about registries, their various purposes and different uses that making an inventory, and standardizing or prescribing good PR design features is a very complicated matter. Nevertheless, efforts toward this end are under way, such as the European recommendations,31 or the Italian27,28 and Spanish RD groups.23

What this study adds

The study obtained agreement on half the questions in the first round and on 89% of questions by the final round. Considering the usual participation rates for online health-related questionnaires, these are very adequate results.32,33 They may have been due to the use of online tools, which improved response time and allowed participants to answer from any location. Participation exceeded 50% in all three rounds, with the highest rate being attained in the last round. The number of participants seems appropriate; it was high enough to provide a variety of opinions while still limited enough to minimize the risk of noise. In addition, professionals came from an optimal range of backgrounds, which translated into a wide variety of points of view.

There was some initial confusion surrounding the terms ‘PR’ and ‘disease registry’, but agreement was eventually reached on a definition of PR. The definition adopted was that provided by the Agency for Healthcare Research and Quality,13 which is the most widely accepted one.4,25,26

In the opinion of the experts consulted, a registry should help promote and enhance clinical research, and it should always be created using well-established quality criteria. The conclusions from the workshop on PRs for RDs34 suggested that data quality should be ensured by the use of focused data management, and that physicians should be convinced of the importance of a registry. In the same way, the Italian RD

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### Table 3 Answers reaching consensus with single choice

<table>
<thead>
<tr>
<th>Q6</th>
<th>Items</th>
<th>Freq</th>
<th>Percentage</th>
<th>95% CI</th>
</tr>
</thead>
<tbody>
<tr>
<td>Consensus in first round (n = 25)</td>
<td></td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>4</td>
<td><strong>Fundamental purpose of a registry</strong></td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td></td>
<td>Study the natural history of disease</td>
<td>8</td>
<td>32</td>
<td>14.6–48.0</td>
</tr>
<tr>
<td></td>
<td>Assess the safety, effectiveness and quality of medical treatment</td>
<td>7</td>
<td>28</td>
<td>11.7–43.7</td>
</tr>
<tr>
<td>6</td>
<td><strong>Preferred geographic area</strong></td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td></td>
<td>National</td>
<td>14</td>
<td>56</td>
<td>33.7–70.0</td>
</tr>
<tr>
<td>Consensus in second round (n = 24)</td>
<td></td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>3</td>
<td><strong>Most important characteristic of a disease for it to be included in an RD registry</strong></td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td></td>
<td>Disability and quality of life (hospitalization, interventions etc)</td>
<td>12</td>
<td>50</td>
<td>28.1–64.7</td>
</tr>
<tr>
<td>5</td>
<td><strong>Type of population that should be included in a registry</strong></td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td></td>
<td>Reference population (healthcare or other)</td>
<td>12</td>
<td>50</td>
<td>31.0–69.0</td>
</tr>
<tr>
<td></td>
<td>Management of a reference population (as census)</td>
<td>12</td>
<td>50</td>
<td>31.0–69.0</td>
</tr>
<tr>
<td>7</td>
<td><strong>RDs that are most needed in a PR</strong></td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td></td>
<td>a. More than 0.1/10 000 (ex Charcot-Marie)</td>
<td>12</td>
<td>50</td>
<td>30.2–67.9</td>
</tr>
<tr>
<td></td>
<td>b. With specific treatment (orphan drug)</td>
<td>16</td>
<td>67</td>
<td>46.4–82.3</td>
</tr>
<tr>
<td></td>
<td>c. Endocrine and metabolic and nervous system diseases</td>
<td>19</td>
<td>79</td>
<td>50.4–83.5</td>
</tr>
<tr>
<td>17</td>
<td><strong>Form in which data should be transferred</strong></td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td></td>
<td>Anonymized: identification data encoded or no data</td>
<td>12</td>
<td>50</td>
<td>29.4–66.8</td>
</tr>
</tbody>
</table>

**Note:** The two highest scoring responses for each question are shown.

**Question.**

**Absolute frequencies.**

**Bayesian credible intervals.**

**Number of participants.**
registry performed an assessment of data-collection completeness and consistency to improve the quality of results. Experts also stated that the two main objectives of a registry are to ‘study the natural history of the disease’ and to ‘assess the safety, effectiveness and quality of medical treatment’, with no agreement on which is the more important one. This lack of consensus suggests that there may be more plausible options, and the literature does, indeed, show very different possible aims for an RD registry. A questionnaire on the current situation of RD registries indicates that ‘epidemiological and clinical research’ is the main aim of registries in Europe, a result in line with the recommendations of the EU Committee of Experts on RDs (EUCERD). The national scope was considered the best option for an RD registry. In 2005, only 29% of healthcare registries in Spain covered the whole of the nation, while currently 61% of registries in Europe are national. Data are usually collected in regional areas but information is then transferred to a national registry, such as the Italian RD Registry. The EUCERD recommendations do not specify what area a registry should cover, but data collections need to be as much internationally interoperable as possible. Also, in line with the literature, RD registries should be managed by national health authorities and supervised by public-health technicians or epidemiologists.

Results showed that experts thought that the common dataset of a registry should contain patients’ ID numbers and dates of birth. These results coincide with other authors’ proposals. Experts also concluded that the inclusion of a specific RD in a registry should be based on its prevalence (>0.1/10,000), the availability of orphan drugs for its treatment, and the disease being one of the endocrine, metabolic or nervous system.

The experts agreed on the need to request patients’ informed consent and to anonymize all data. Similarly, the EUCERD recommendations require informed consent and state that patients’ identities should be kept as confidential and secure as possible. Currently, only 10.8% of European registries use irreversible anonymization; 47.6% use reversible anonymizing and 41.5% use identifiable data. In addition, 33% of registries do not request patients’ informed consent. More efforts should, therefore, be made to improve data protection.

Our study identified two items that experts disagreed on. The first item was the most important characteristic of a disease for it to be included in an RD registry (question 3), which experts first identified as ‘disability and quality of life’, but by the final round there was no longer a consensus; a possible explanation for this could be that it is difficult to choose a single item that determines whether a disease should be

| Table 4 | Answers reaching consensus with a graduation response$^a$ |
|---|---|---|
| Q$^b$ | Items; consensus in second round (n$^c$ = 24) | Mean | $SD^d$ | 95% CI$^e$ |
| 1 | Definition of PR (1–5 scale) | ‘Organized system that uses observational study methods to collect uniform data (clinical and other) to evaluate specific outcomes in a population defined by a particular disease, condition, or exposure, and that serves a predetermined purpose, whether scientific, clinical or political’ | 4.2 | 0.9 | 3.8–4.6 |
| 13 | Advantages of a PR (1–5 scale) | ‘Health information system constituted by a file of documents that contain uniform individual information and which is collected in a comprehensive and systematic way to meet a predetermined purpose’ | 3.7 | 1.0 | 3.3–4.2 |
| 18 | Registry information that should be made public (1–10 scale) | Serves as a tool to locate or identify cases for researchers and patients interested in participating in studies or in clinical or therapeutic trials. | 4.0 | 1.0 | 3.6–4.5 |
|  | | Connects registries with other databases to provide a complete overview of the disease and to obtain additional results | 3.3 | 1.4 | 2.7–3.9 |
|  | Lethality | 6.7 | 3.0 | 5.4–8.0 |
|  | Frequency of pathology | 6.5 | 3.5 | 5.0–8.0 |

$^a$The two highest scoring responses for each question are shown. $^b$Question. $^c$Number of participants. $^d$Standard deviation. $^e$Bayesian credible intervals.
included in a registry. The second source of disagreement was the type of registry information that should be made public, probably because most experts considered it important to disseminate all available information.

Limitations of this study

This study on RD was conducted at the national level, with all the inherent limitations; a supranational study would very probably be of greater interest. Concerning the Delphi method used, it provides a specific view of the study’s subject matter and could be useful for implementing measures to improve rare-disease PRs. There is, however, no cut-off point for the level of consensus achieved. The literature sets the cut-off point at somewhere between 50 and 80% agreement. Since this is the first consensus study on RD characteristics for a PR, we took our own decision to set the cut-off point at 50%. We aimed to identify the most important characteristics and not focus on a single item for each question. The Delphi method also has an obvious inherent limitation: results are highly dependent on panel composition. We, therefore, endeavoured to gather a heterogeneous and multidisciplinary expert group with representatives from various kinds of institutions and organizations in an anonymous process that made it impossible to identify the characteristics of the experts participating in each round.

Supplementary data

Supplementary data are available at PUBMED online.

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