2018 User Satisfaction Survey
of the Orphanet Website

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A. METHODOLOGY

This year the Orphanet satisfaction survey has been totally rethought in order to be more direct and effective in terms of tracking improvements. This year's satisfaction survey has been conducted in two phases:

- Phase 1 was launched in January 2018: a popup window was added to the first page users landed on. The survey was translated into the 8 languages of the website (i.e. English, French, Spanish, Italian, Portuguese, Dutch, German or Polish) and was displayed in the language of consultation via a pop-up. The survey was closed after 3 weeks of display on the website. A survey was made of 12 questions split into 4 sections.
- Phase 2 was a consultation of respondents in Phase 1 who declared that they were willing to give more insight concerning their opinion on specific products. This second phase started in mid-March and ended on 1 April 2019. The survey was translated into the 8 languages of the website and was sent respecting the language in which the users answered the first part of the survey.

The results from all of the languages of the survey were consolidated and then analysed. The results of this analysis are presented in this report with elements of comparison as regards last year's results. The number of total respondents for each question are given. Part B of this document presents the results of Phase 1 of the survey, while Part C & D presents the results of Phase 2.

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

B. RESULTS OF THE PHASE 1 OF THE SURVEY

This on-line survey was designed in November 2018, using the online survey tool Survey Monkey. Questions focused on the professional occupation of the users, their habits when they visit the Orphanet website, their opinion of the content as well as their overall satisfaction and their suggestions for improvement. Replies were mandatory for all questions, apart from questions 7 and 9 which are designed to allow respondents to give detailed feedback if they wish. A total of 10086 users provided answers.

I. Part 1: Methods and reasons for using the website?

Question 1: How did you discover Orphanet?
This question aimed to determine how respondents first learnt about Orphanet. Only one choice was possible. 10086 respondents replied to this question.

A large majority of our users discovered Orphanet via Google (69.9%, figure 1). Word of mouth has also brought a significant percentage of respondents to Orphanet (around 14%). The other vectors cited by users include websites of patient organisations or hospitals, training sessions/lessons, or rare disease related events. Compared to last year, these results show a significant increase of access through Google (nearly 16 points) while word of mouth falls by 11 %. The remaining vectors are marginal this year compared to the first two vectors. These results are the same in all the languages investigated.
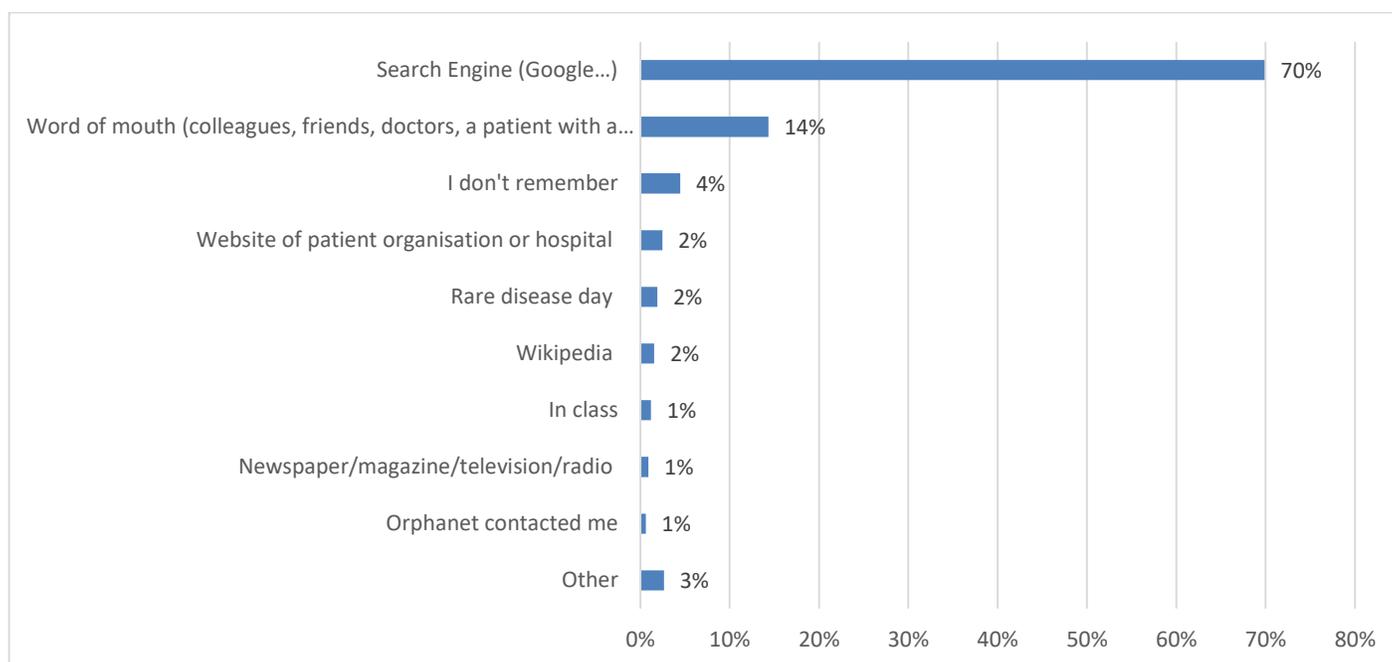
Figure 1: Mode of discovery of Orphanet by respondents (n=10086).

Question 2: What sort of information are you looking for during THIS CONNECTION to Orphanet?

This question aims to determine which kind of information visitors sought on Orphanet. Unlike last year, it was not possible to select several categories. This may explain the difference in results compared to last year. Moreover, this change bothered several respondents. Next year, we will allow respondents to choose multiple categories again. 8321 respondents replied to this question.

This year results show the same top three consulted products as last year's survey (figure 2): information for a specific disease (57.7%), information on rare diseases in general or on the Orphanet project (22.8%), and clinical guidelines (3.9%). Our visitors also look for information on rare disease nomenclature / codes (2.8%), specialist clinics/centre of expertise (2%), classifications (1.6%) and diagnostic tests (1.6%)

What sort of information are you looking for during THIS CONNECTION to Orphanet?

Category	Percentage
a specific disease	58%
rare disease in general, the Orphanet project	23%
clinical guidelines	4%
rare disease nomenclature/ codes	3%
specialist clinics/ centre of expertise	2%
classification	2%
diagnostic tests	2%
gene	1%
disabilities	1%
reasearch projects	1%
patient organisations	1%
clinical trials	1%
epidemiology	0%
Orphan drug (a specific one or in general)	0%
datasets for research (Orphadata)	0%
newsletter	0%
visit to register your activity in Orphanet	0%
visit to contribute to Orphanet	0%
registries	0%
other (please specify)	1%

Figure 2 : Information sought by respondents during their connection to Orphanet (percentage of total number of respondents n=8321)

Question 3: How often do you visit Orphanet?

Only one response was possible. 8097 respondents replied to this question. Around 12% of those answering the survey are regular users, whereas 44% were visiting Orphanet for the first time, the remaining 44% visited either over twice a month, or over twice a year.

There were 17% (figure 3) more first time users this year compared to last year (27%). On the other hand, there was a decrease in users consulting the site more than twice a month (33% compared to 23%), and a slight decrease in the number of respondents stating they use the site more than twice a week (17% compared to 12%).

This trend is the same as last year's and shows that people tend not to come back to Orphanet once they find the information they want or that first time users are more prone to answer to this survey as regular users already did last year or before.
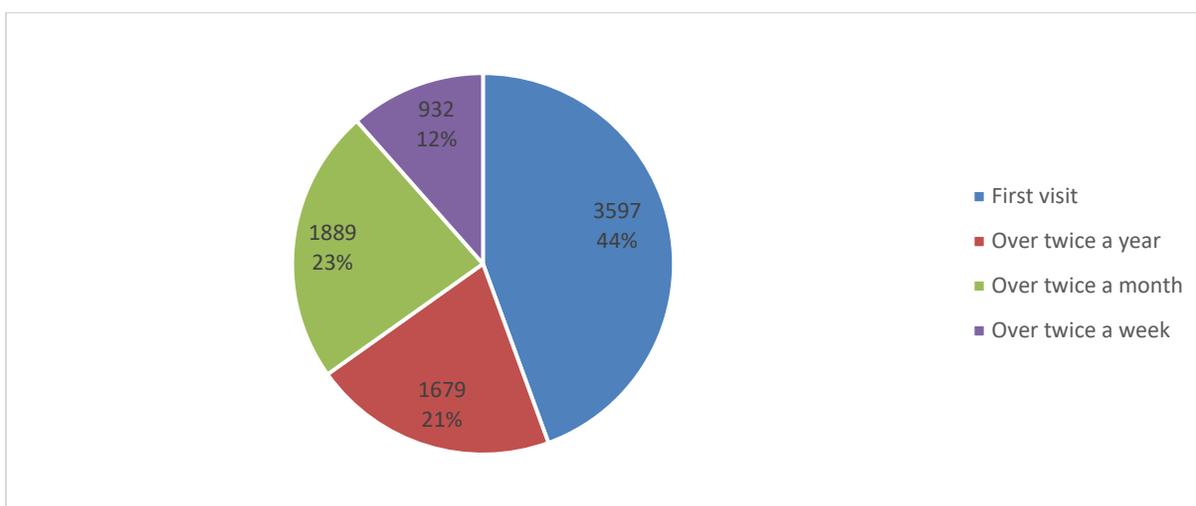
Figure 3 : Visiting frequency of respondents (n=8097)

Question 4: How are you accessing Orphanet today?

This question was aimed at finding out more about the type of hardware used to access the site. Only one response was possible for this question. 8097 respondents replied to this question
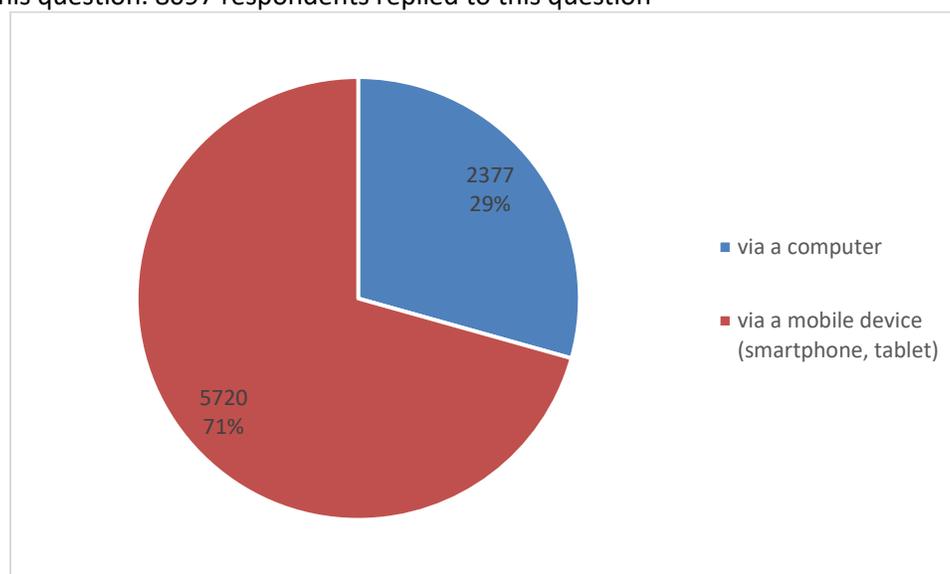
Figure 4: Mode of accessing Orphanet by respondents (n=8097)

Of those who responded to the survey, 71% did so using a mobile device with an Internet connection (figure 4). This represents a progression of 27% compared to the previous year's survey and confirms the trend seen last year, namely that more and more people use a mobile device to connect to the Orphanet website.

II. Part 2: Your opinion on Orphanet's products/ services and its website

This part of the questionnaire was aimed at deciphering our users' global satisfaction about our website and our products/services. This part provided also the opportunity to respondents to tell us what we should improve or

develop to better serve their needs. In this part, we also asked the respondents if they agreed to be contacted to give us amore detailed opinion on the product(s) of their choice(s).

Question 5: To what extent do you agree or disagree with the following statement?

This question aimed to find out whether users found Orphanet website user friendly, easy to use and if the information found was easily understandable. First time users were asked this question due to a technical issue. However, their responses were not taken into account, as they had no established experience of the site before answering to the survey. According to their experience on the website, respondents were asked to rate (++, +, -, --), to what degree:
- the site is easy to navigate,
- the information was easy to find,
- the information found was easy to read and understand.

Respondents were asked to answer to the three items. 4724 answers were taken into account to this question, corresponding to all the answers given by the non-first time user.
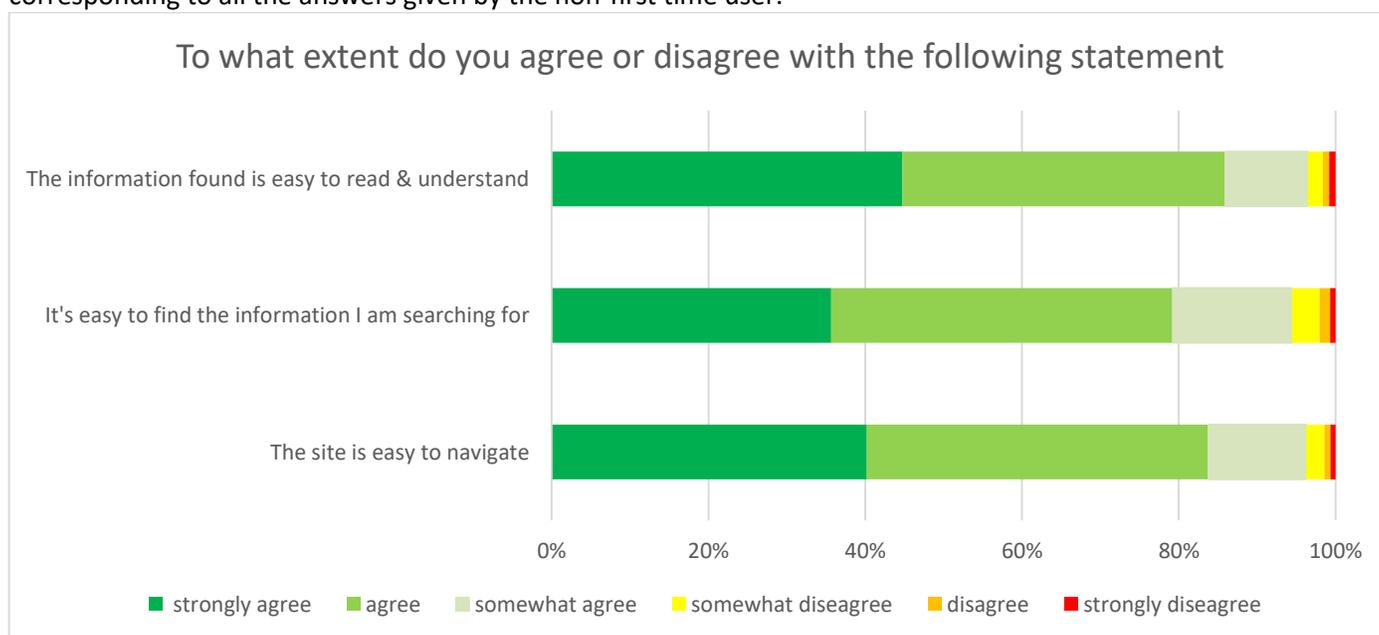
Figure 5 : Repartition of the respondents rate on the ease to use the Orphanet website and to understand the information found (n=4724)

These results show that most of the respondents (~ 80%) find that the Orphanet website is user-friendly, that the information is easy to find and that once it is found the information is easy to read and understand (figure 5). A very small minority of respondents (~2%) disagree or strongly disagree with these statements. Depending on the statement 13 to 19% of respondents are ambivalent. These results should, however, be related to question 9. In this question, we asked what Orphanet could do to better serve its users. Answers to this question showed that users don't know certain Orphanet functionalities or services and that Orphanet must make further efforts to make its site even easier to use and clearer.

Question 6: Overall, how satisfied are you with Orphanet?

The global satisfaction of Orphanet website users was evaluated through this question. First time users were asked this question due to a technical issue. However, their responses were not taken into account, as they had no established experience of the site before answering to the survey. Only one response was possible between Very

satisfied, satisfied, dissatisfied and very dissatisfied. 4369 answers were taken into account to this question corresponding to all the answers given by the non-first time user (figure 6).

The vast majority of respondent were either very satisfied or satisfied with Orphanet with a total of 93% responding positively. 6% were dissatisfied and 1% very dissatisfied.
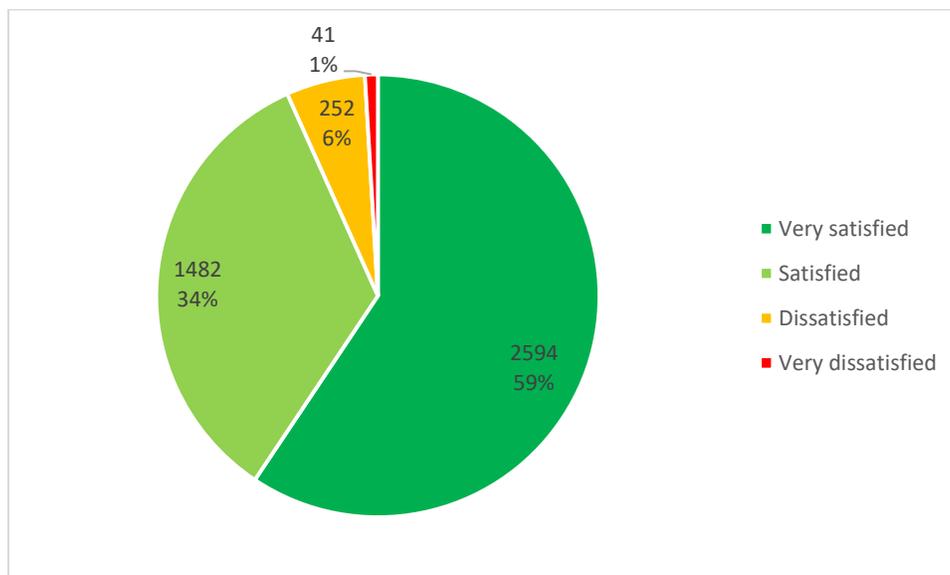
Figure 6: Global satisfaction of the Orphanet website users

Question 7: Which of the following words would you use to describe Orphanet ?

This question was aimed at ascertaining in more detail what people think of Orphanet. First time users were asked this question due to a technical issue. However, their responses were not taken into account, as they had no established experience of the site before answering the survey. Respondents could choose as many choices they wanted in the following list: reliable/unreliable, high quality/poor quality, useful/ impractical, unique/ ineffective, up to date /not up to date. People were not obliged to answer this question. 4500 persons responded to this question and selected a total of 9654 adjectives.

Results show that respondents are quite happy with Orphanet (figure 7). Usefulness, reliability and high quality of this website are the main characteristics that people retained with 31%, 28% and 20% of responses respectively. The fourth most used adjective is « up to date ». This result is however in contradiction with one of the most common suggestions made to question 9, where respondents requested that Orphanetbe more up to date.
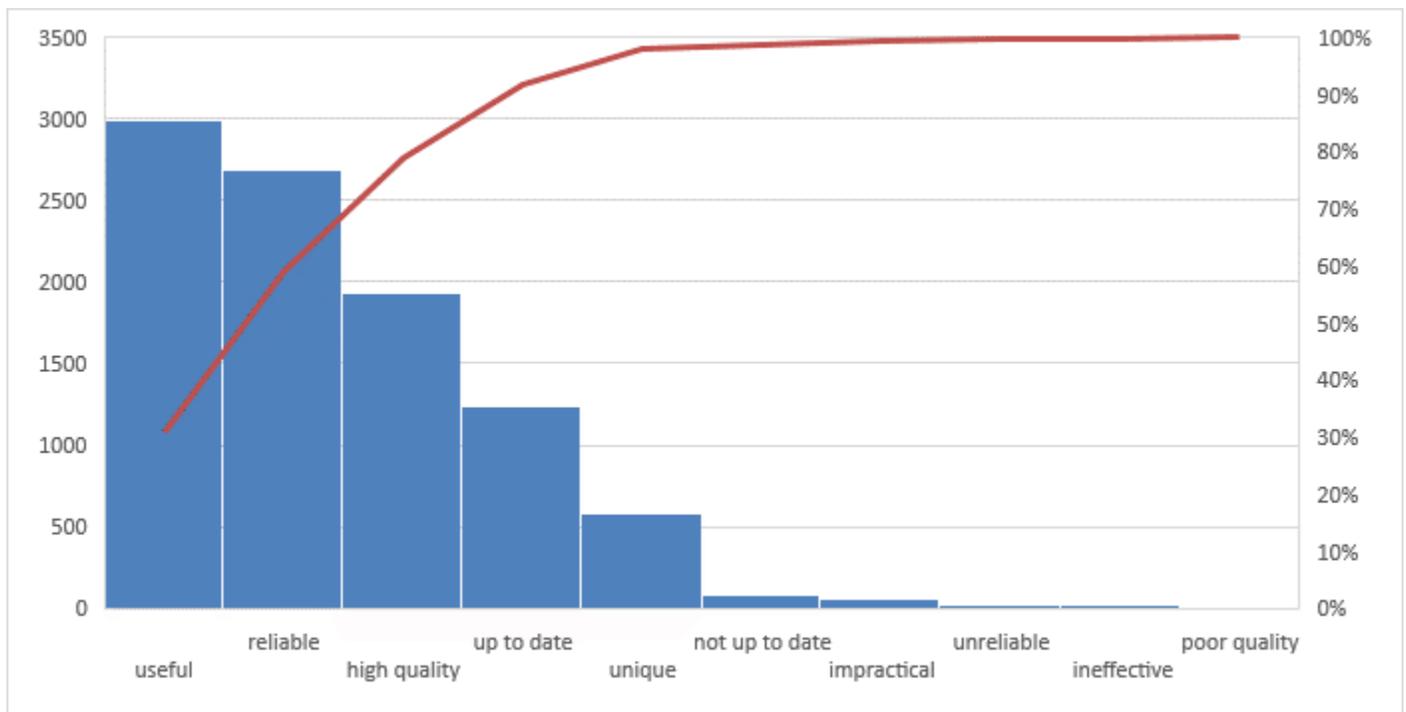
Figure 7: Pareto graphic representing which of the following words the respondents would use to describe Orphanet

## Question 8: How likely is it that you would recommend Orphanet to a friend or colleague?

This question was asked in order to determine Orphanet's Net Promotor Score (NPS), which measures the likelihood, on a 1-10 scale, that someone will recommend a company to someone else. Net Promoter Score was calculated by subtracting the percentage of customers having ranked this probability from 0 to 6 from the percentage of customers having ranked this probability from 9 to 10. Thus, results can vary from -100 to +100.
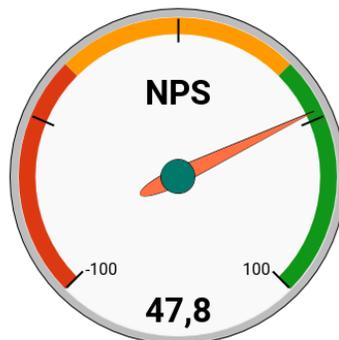
Figure 8: Net promotor score (NPS) of Orphanet according to the annual satisfaction survey of 2018 is 47.8

Out of the 4199 responses collected, 2409 turned out to be promoters as they responded with a score of 9 or 10, 1389 were passive (score of 7 or 8) and 401 were detractors (score of 0 to 6). This gives a NPS of 47.8 (figure 8).

To our knowledge, Orphanet is the only service dedicated to providing free information on rare diseases and orphan drugs that publishes its NPS. Thus, we cannot compare this score to other similar services and this score must be considered as a starting point in measuring our customer satisfaction that we will track over time. However, it is noteworthy that according to these results ~60% of the respondents would recommend using Orphanet while only 10% would not.

Question 9: What should Orphanet do to better serve your needs?

Comments were provided by 1187 individuals in response to this question. First time users, as they have no established experience of the site, were not asked this question.

Around 20% of those who provided comments were satisfied by the service provided by Orphanet, or left messages of encouragement and support concerning Orphanet's activities. The Orphanet website was reworked in March 2017 to improve its user friendliness. Despite this total make over, around 15% of the collected responses showed that the website in not user friendly enough. Indeed, among these comments, around one third were to ask for services or products that are already provided by Orphanet but that are apparently not visible enough on our website. Many comments also concerned the search engine that was judged to not be sufficiently efficient. Many users also highlighted the need to improve the search functionalities and the search engine: the IT infrastructure is still being overhauled and hopefully this will improve in the near future. Comments also gave a number of suggestions on how to improve the presentation of the data in this complex database, notably in improving the readability of the disease summary text or in highlighting the resources linked to each disease. Users were also interested in additional help on how to best use the Orphanet website either through tutorials or via additional information on how to navigate and access definitions of concepts used in the website.

Another Orphanet's users main request was that Orphanet more frequently update its data. Concerning the update of disease summaries, Orphanet is currently actively working on this matter and is changing its update strategy in order to be able to update a significantly greater number of abstracts per year. Notably from now on, some disease summaries will be directly produced by European Reference Networks (ERNs)[1] that agreed to work with Orphanet. This strategy has been launched at the end of the first semester 2019 and it is hoped that it will enable us to improve the update frequency of the abstracts.

Concerning the updates of the expert resources (expert centres, patient organisations, medical laboratories and their associated diagnostics tests, research projects, clinical trials and registries), Orphanet is very dependent on the good will of the professionals as they are the only ones that can update this data. This is why Orphanet carries out an annual email campaign to professionals and asks them to update their data. In parallel, professionals are able to update their data via the Orphanet professionals' registration tool at any time throughout the year.

Another regular comment (~10% of respondents) was that the disease summaries be more detailed in the presentation of the disease, the treatment, the diagnostic criteria.

Users were also interested in receiving notification of updates to certain types of data, or concerning specific diseases. Users also requested that more information be made available in the different "Orphanet' languages"[2] and that information be made available in even more languages, so that patients and healthcare professionals can access this information in their own language. This is dependent on the availability of national funding for the translation; in 2018 the site has been made available in Polish.

Many respondents have suggested that photos, or medical imagery, could be added to aid diagnosis. Some comments concerned the reintroduction of the aid-to-diagnosis tool (Orphanet-Phenomizer tool) that Orphanet took off-line following feedback from users has this tool was not efficient. Orphanet is however, testing a new aid-to-diagnosis tool in order to include it in Orphanet.

Users also suggested improvements such as the introduction of a dynamic glossary to explain medical terms for non-professional users to make the site understandable for the general public and to provide links to ongoing research

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[1] 1 European reference network (ERN) is a network connecting health care providers and centres of expertise of highly specialised healthcare, for the purpose of improving access to diagnosis, treatment and the provision of high-quality healthcare for patients with conditions requiring a particular concentration of resources or expertise no matter where they are in Europe.

[2] That is to say French, English, German, Spanish, Dutch, German, Italian & Polish

papers when an Orphanet text is not available or in provide additional information. It should be noted that Orphanet already provides links to publications, including for general public, as well as for clinical practice guidelines and links to PubMed: this is perhaps not visible enough to our users

A number of comments highlighted the need to update the Orphanet mobile app and to fix several bugs: work has been carried out in the past year to improve and update this app and it is hoped that it will be launched in 2019.

Some comments highlighted that resources could be better known by the general public and health professionals, in particular general practitioners. One of the main focuses of the Orphanet operating committee is to explored ways to improve national outreach. We hope this will allow Orphanet to progress on this matter.

III.   Part 3: Orphanet Application

Question 10: Have you downloaded the Orphanet application? If not, why have you not downloaded the app? If yes, are you happy with the app?

Only one response was possible for this answer. Around 7% of respondents had downloaded the Orphanet mobile app. This figure is the same as last year. Those that have not downloaded the app (93% of respondents) were asked the reason why they have not downloaded it (figure 9) and the ones that did were asked if they were satisfied with it.
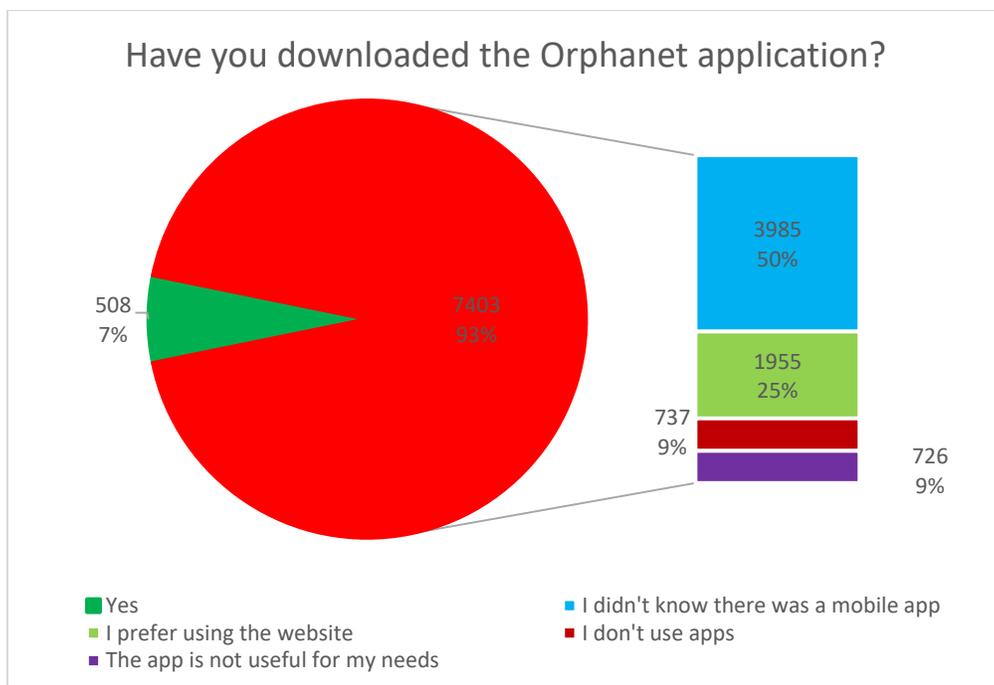
Figure 9: Respondents not having downloaded the Orphanet mobile application (n=7403) and reason why they didn't

The principal reason (50%) for not downloading the app was that users did not know it existed. Around a quarter stated that they prefer using a website to an app, and 9% stated they do not use apps in general with 9% stating that the app was not suited to their needs. For these last three categories, the repartition is the same as last year. However for the first one ("users did not know the app existed"), this represents a decrease of 15% compared to last year and this shows that efforts Orphanet made to promote this tool had some impact (for instance, information about the app and a link to download it appeared next to this question in the survey). According to these results, Orphanet should look into the matter further in order to decipher if this application should be maintained as it is and

make more efforts to promote this tool or if it has no added value compared to the Orphanet website and should be shut down in order to create a new one dedicated to a more specific audience.
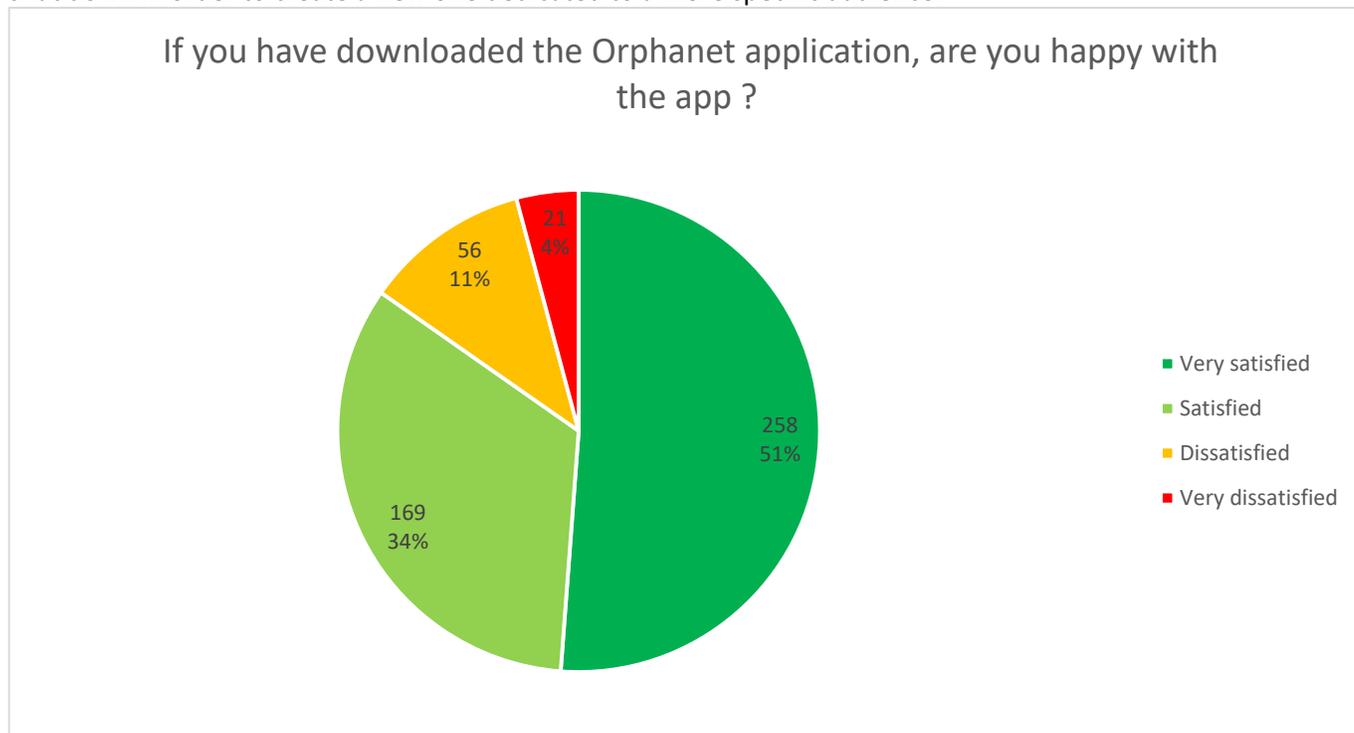
Figure 10: Satisfaction of the respondents having downloaded the Orphanet app (n=508)

Results showed that the vast majority of people that have downloaded the Orphanet application (85%) are either very satisfied (51%) or satisfied (34%) while 11% of respondents are not satisfied and 4% are very dissatisfied (figure 10). In order to better understand why they are not satisfied we should allow them to explain why in next year's survey.

IV.   Part 4: More information about you

Question 11: What country do you live in/work in?

This question was aimed at ascertaining the geographical location of users replying to the survey. Only one response in a drop down menu was possible for this question. This menu contained the 250 countries of the ISO norm 3166-1 alpha-2. 6436 replies were registered for this question. The top ten countries replying to the survey were (figure 11): Italy (n=1345), France (n=1286), Brazil (n=486), Mexico (n=375), Spain (n=358), Germany (n=285), United States of America (n=271), Argentina (n=176), Colombia (n=157) and Belgium (n=105).

This is the same trend as last year as eight of the top 10 countries were also in the top ten of last year. Only Argentina and Colombia were not present in last year's results while Canada and Switzerland were.
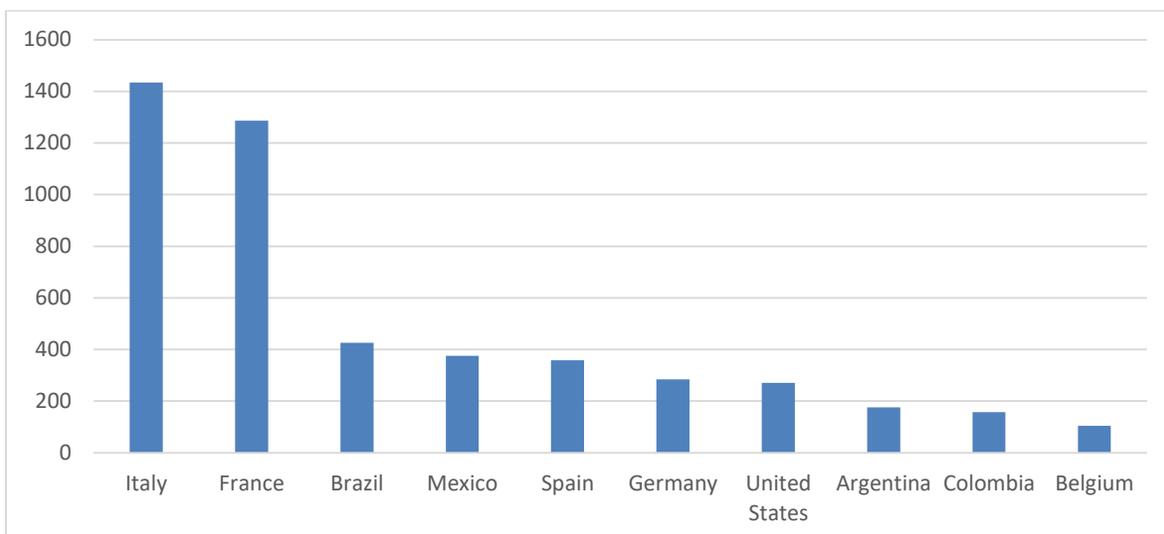
Figure 11 : Top 10 countries replying to the annual satisfaction survey

Question 12: In what capacity are you consulting the Orphanet website TODAY?

This question aimed to determine the profile of Orphanet's users. Seven categories were proposed (i.e. health professional, patients/entourage, researcher, industry, health care manager/policy maker, education/communication and students), and a free text field was included for other types of users to enter their profession. Only one response was possible. Respondents from the 'other' category were reassigned to one of the seven proposed categories when appropriate. 6436 replies were registered for this question.

Figure 12 shows the distribution of respondents amongst these categories:
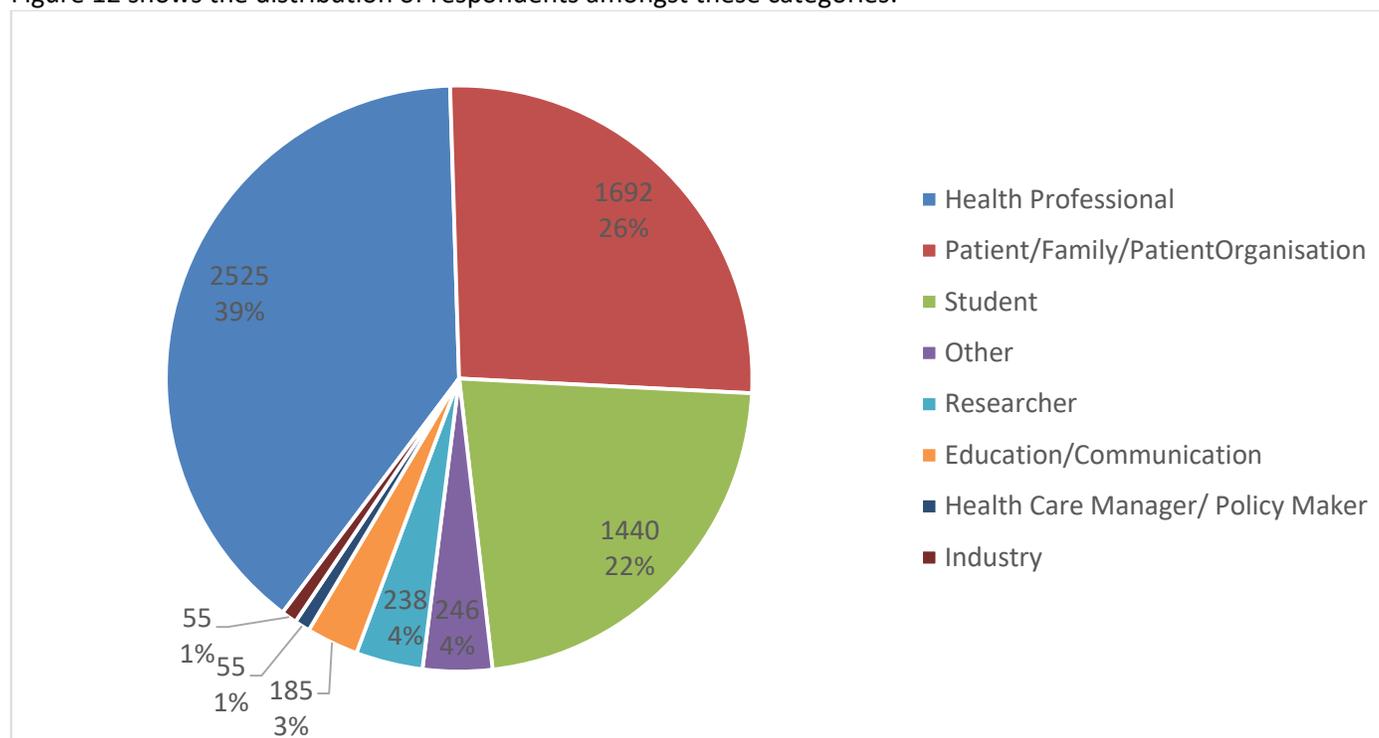
Figure 12 : Types of Orphanet user (number of responses and percentage of total responses). N =6436

The largest category of respondents is the health professional category (39%). The second largest category of respondents is patients and their entourage (including patient organisations, alliances and support groups) with 26% of responses. Many students (22%) also replied to the survey. More than half of the 'other' category was made of

persons that describe themselves as just 'curious' (n=150). The rest of this category was, for the majority, respondents that did not state their professional category (n= 58)

This year there was a slight decrease in the percentage of health professionals responding to the survey (39% this year, 46% last year), and a slight increase in the percentage of students responding to the survey (22% this year,18% last year). The percentage of patients and their entourage remained stable.

Then, for each category, respondents were asked to choose the sub-category that would best describe them. If they answered 'other' they were invited to state in which capacity they were answering. Only one response was possible. Respondents from the 'other' category were reassigned when appropriate

This year all respondents were also asked if they had an expertise in the field of rare diseases. 43% responded that they had expertise in this field. These results are similar to those of last year even if last year we only asked this question to respondents that declared to consult Orphanet as health professional.
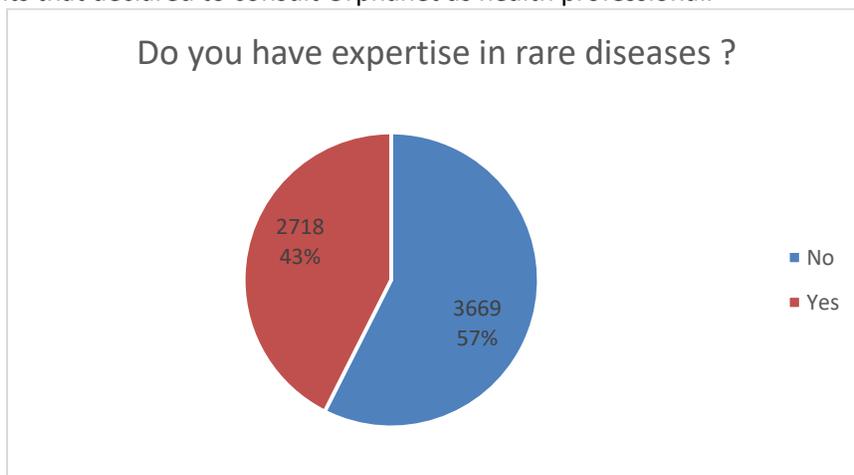
Figure 13 : Repartition of respondents considering having a expertise in rare disease.

Health professionals (n= 2492):

Hospital or independent specialists represent by far the main category of respondents (54%). Altogether, 71% are medical doctors (figure 14).

11% of the respondents are healthcare professionals other than medical doctors and nurses (e.g. psychologists, physiotherapists, dentists etc...), which represents an increase of almost 6% compared to last year's survey.

Other professional categories remain equally represented compared to last year.
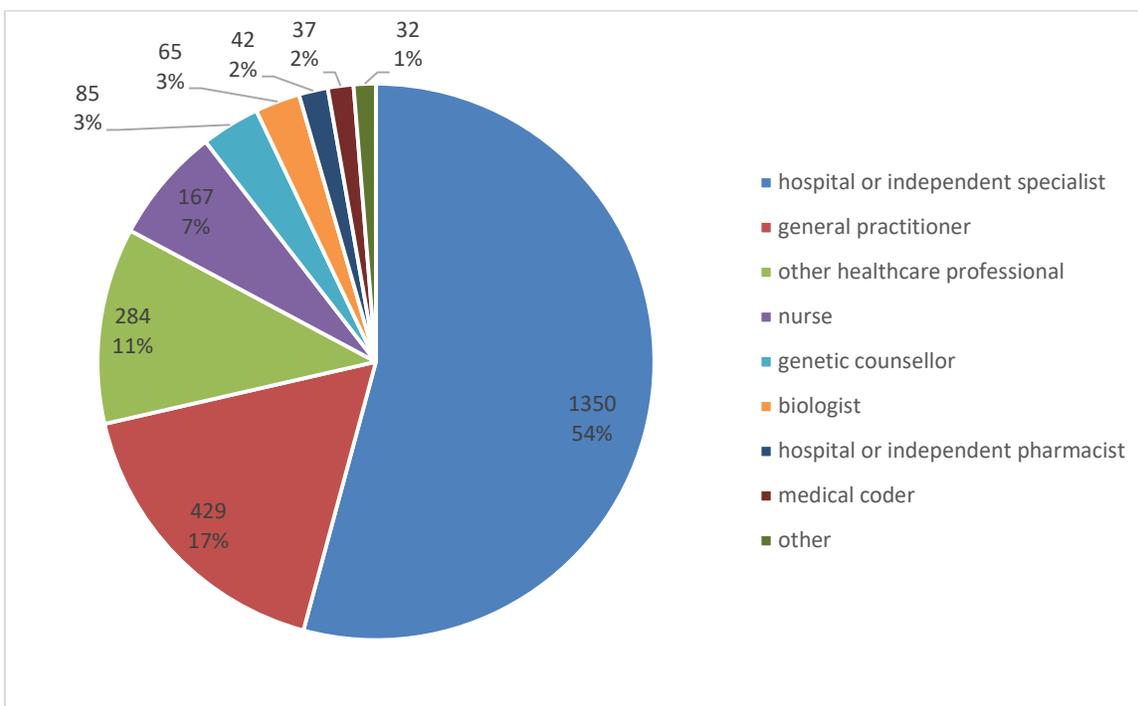
Figure 14 : Types of respondents qualifying themselves as health professionals

Patient/entourage (n= 1673):

Most of the people who selected this category are patients (48%, figure 15); 42% are family members of a patient with a rare disease. The results are globally similar to those of the previous year.
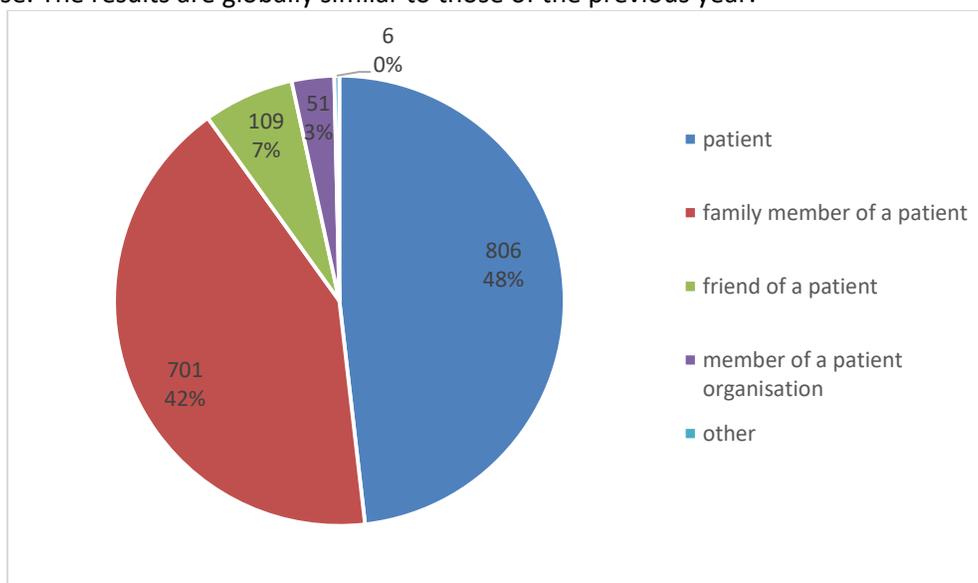
Figure 15 : Types of respondents qualifying themselves as a patient or part of a patient's entourage.

Researcher (n= 236):

Academic researchers represent 56% of respondents of the research category (figure 16). The percentage of social sciences researchers that responded to this survey slightly increased compared to last year (12%, up from 8.3%), as did the percentage of bioinformaticians/ biostatisticians (9% this year, 5.1% last year).
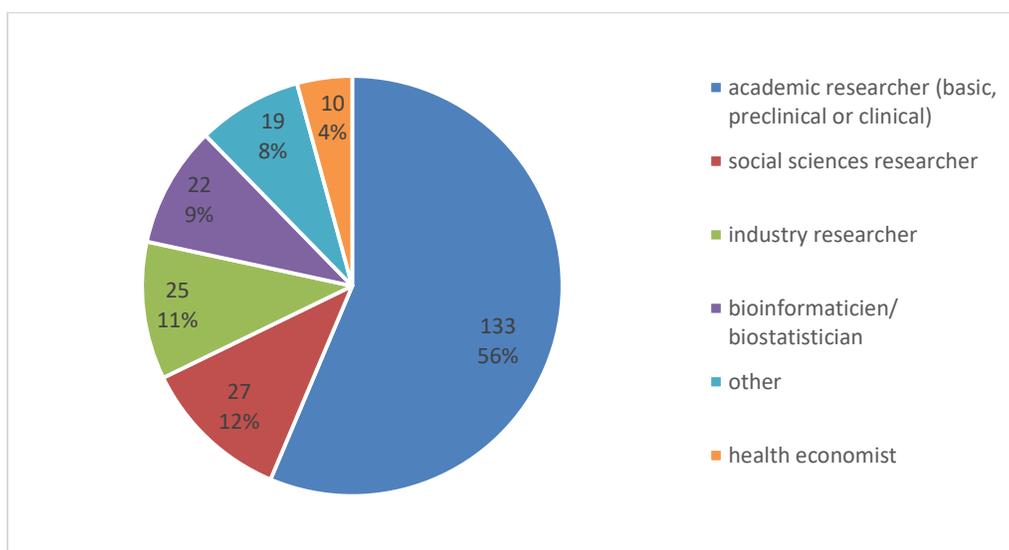
Figure 16: Types of respondents qualifying themselves as working in the field of research.

Industry (n=57):

45% of respondents in this category this year are consultants in the sector while 37% work in the biotechnology or pharmaceutical industry (figure 17). Compared to last year, the percentage of respondents working in the biotechnology and pharmaceutical arena dropped by 23 % while the percentage of consultants in industry went up by 26 %, however the small sample size (57 this year) should be noted when assessing this evolution.
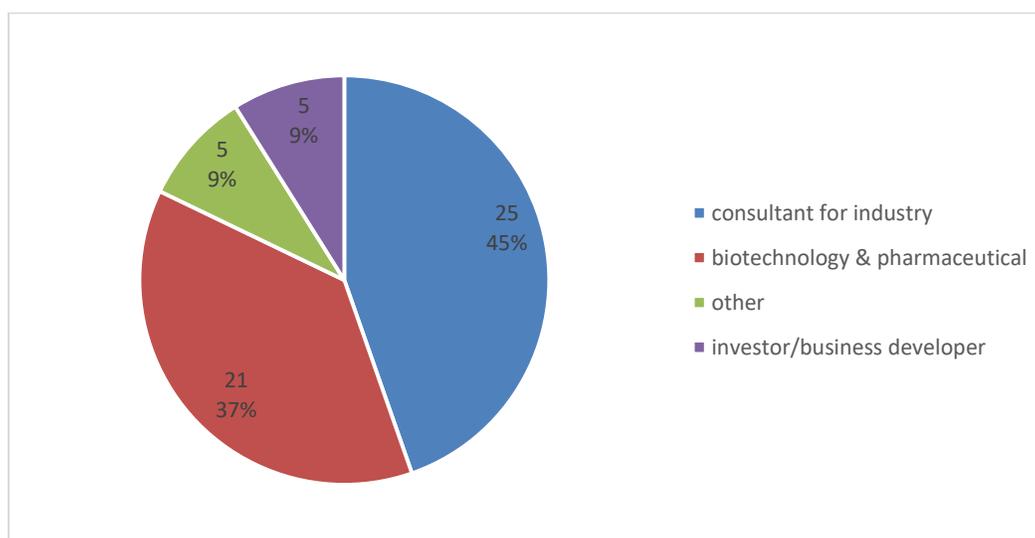
Figure 17: Types of respondents qualifying themselves as working in the biotechnology or pharmaceutical industry

Health care manager/policy maker (n=51):

In this category, 39% work in European administration sector, 10 % in governmental administration and 10 % in hospital administration (figure 18). These results are quite different from last year as there were no responses from the European administration sector last year and this is the main category of respondents this year. The percentage of respondent working in governmental or hospital administration is lower than last year (respectively 10% this year compared to 34.8 & 37 %). However, one should keep in mind the small sample size (51 this year) when assessing this evolution.
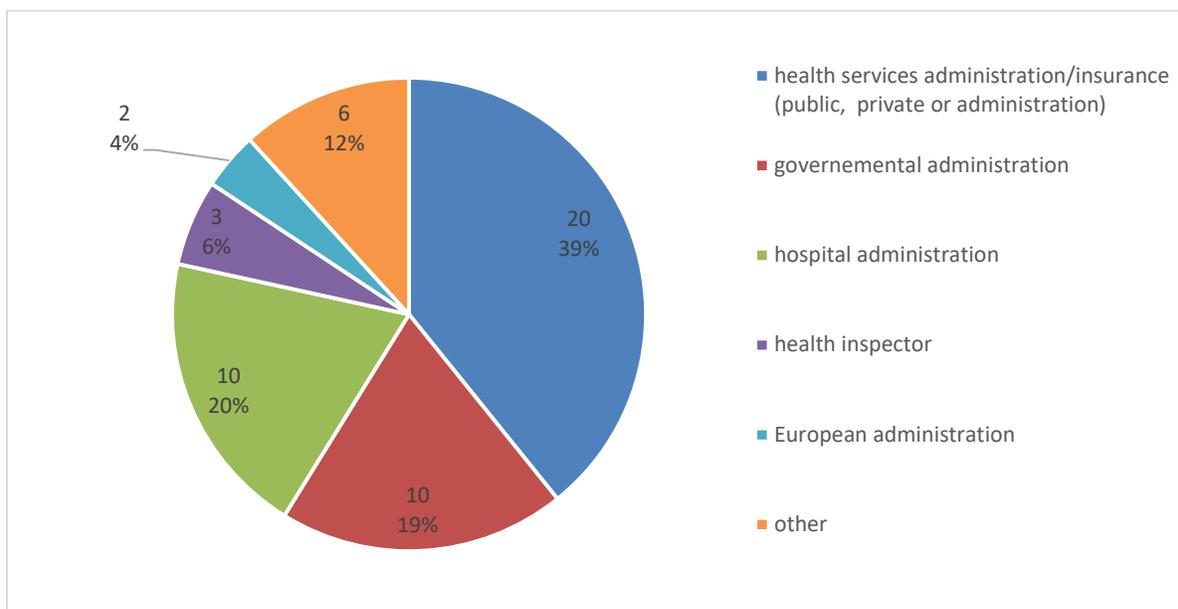
Figure 18: Types of respondents qualifying themselves as working in the field of health care management/policy making.

Education/communication (n=180):

In this category, teachers represented 44% of respondents (figure 19). Social workers were the second most represented sub-category with 13% of respondents for this category. The "other" category included other professionals from the education sector, or professions linked to communication (writer, publisher, information scientist). The results are globally similar to last year.
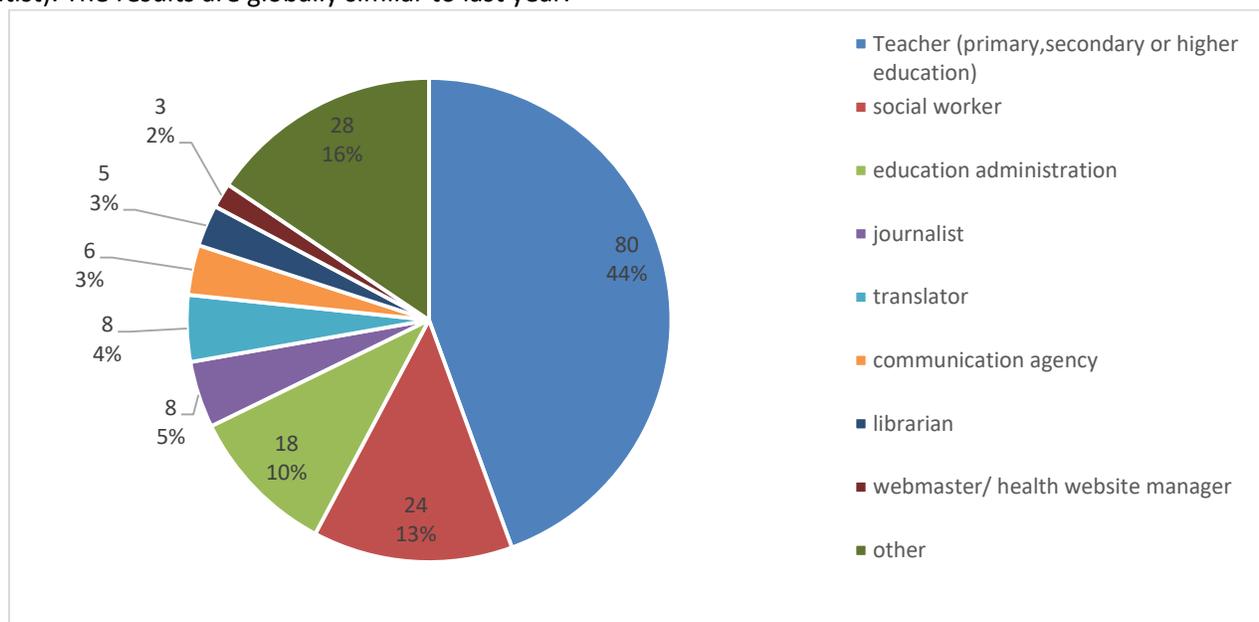
Figure 19: Types of respondents qualifying themselves as working in education/communication.

Students (n=1422):

Medical students represent 76% of this category (figure20). The other respondents were studying in biology, dentistry, genetic counselling, pharmacy, etc. The results are the similar as last year with a small increase of other types of students (24% this year compared to 15.3% last year).
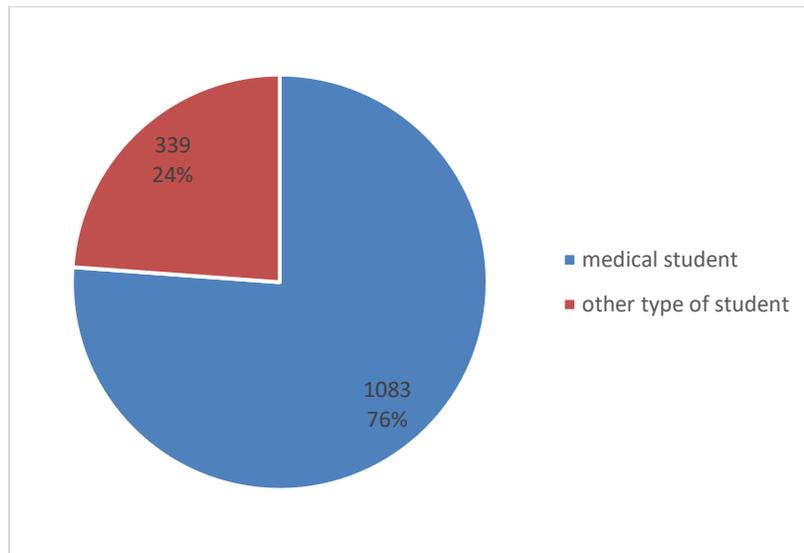
Figure 20: Types of respondents qualifying themselves as students

C. RESULTS OF THE SECOND PHASE OF THE SURVEY: DETAILED OPINION ON THE ORPHANET PRODUCTS

Persons contacted for this phase of the survey were asked to give more insight on their opinion on the following products:

- Orphanet rare disease nomenclature/ codes
- Orphanet Classifications
- Cross-referencing with other reference databases (OMIM, GARD, …)
- Datasets for research (Orphadata)
- Specialist clinics/ centres of expertise
- Patient organisations
- Diagnostic tests
- Registries
- Research projects
- Clinical trials
- Orphan drugs
- Diseases summary texts
- Clinical guidelines
- Epidemiological data
- Natural history of the disease
- Genes
- Disabilities of rare diseases

Respondents could give feedback on as many products as they wanted, knowing that there were 5 to 6 questions for each product. Mandatory replies were depending on the chosen product. Of the 2409 persons who accepted to be contacted, a total of 346 gave answers to at least one product. Taking into account this low rate of answers, the annual satisfaction survey of next year should be rethought in order to be more attractive and allow us to draw conclusion instead of allow seeing trends in the users' satisfaction.

Independent of the number of products they decided to give feedback on, users were asked to describe the capacity in which they use Orphanet. Seven categories were proposed (i.e. health professional, patients/entourage, researcher, industry, health care manager/policy maker, education/communication and students). These categories were identical to the first part of the survey. Note that this was a multiple-choice question and thus users could describe multiple capacities of usage (e.g. health professional and research).

I. <u>Detailed satisfaction survey for the Orphanet rare disease nomenclature/codes</u>

A total of 118 users chose to give their opinion the Orphanet nomenclature.

The most represented response language was Spanish (31%), followed by English (21%), Italian (20%) and French (14%). The majority of respondents identified as health professionals (33%), followed by patients, patient family members or patient organizations (25%) and researchers (16%).

58% of respondents (n= 117) stated that they usually access rare diseases pages through an external search engine (e.g. Google), while 42% use the Orphanet homepage as a starting point to search for a disease. This result is slightly surprising as previous analysis had established that a large majority of users access diseases pages through external search engines (mainly Google).

User experience with the nomenclature on the Orphanet website

Purpose

Respondents were asked why they were using the nomenclature on the Orphanet website. 81% of respondents (N = 115) search for diseases on the Orphanet website in order to obtain information (e.g. clinical, genetic, epidemiological, etc.), while 10% consult the nomenclature for coding purposes. This is in line with the main purpose of the Orphanet website, which is to provide information on rare diseases and associated data and resources. Users involved in rare disease coding are encouraged to use the nomenclature datasets available in Orphadata, as these come in a format that is optimized for coding activities.

Finding the information

Respondents were also asked if they find easily the information they are looking for. Overall, users easily find the disease(s) they are looking for in the Orphanet website (79%, N = 118). Among users who consult the nomenclature for coding purposes (N = 9), a similar proportion agreed that they can easily identify the ORPHAnumbers that they are looking for.

Among the 21% (N=118) of users who stated that they have difficulty finding the disease(s) they are looking for, 40% attributed this difficulty to the diseases nomenclature (main name, synonyms, and keywords) being incomplete or lacking pertinence, while 27% (N=30) concluded that the diseases in question were not present in Orphanet. This difficulty does not seem to be a translation issue, since this reply was selected by as many Spanish speakers as English speakers, and in an even smaller proportion for the other languages.
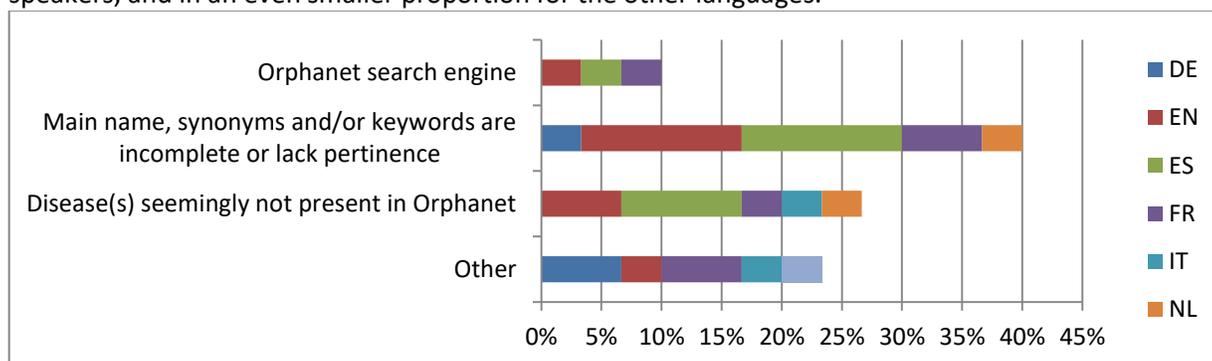
Figure 21 : Difficulties encountered when consulting the Orphanet nomenclature through the Orphanet website, with proportion of each language of response (DE = German; EN = English; ES = Spanish; FR = French; IT = Italian; NL = Dutch).

Among the 7 users who selected "Other", one commented that "the sometimes many synonymous or parallel existing names of a disease complicate the search". This reflects a recurrent complaint that the list of results that is displayed when using the diseases search engine currently does not indicate the level of classification of each ORPHAnumber or the hierarchical relationship between ORPHAnumbers.

The level of classification of each clinical entity is now indicated on its respective disease page since July 2019 (after the survey was conducted), with a direct link to the classifications, as well as in the search result list. Future surveys will help determine the impact of these new features on the users' experience searching for rare diseases on the Orphanet website.

Contributing to the nomenclature

People were asked if they knew they could contribute to the Orphanet nomenclature. 89% of respondents do not know how one can contribute to the Orphanet nomenclature, which can be done through the "Suggest an update" button located on each disease page. The proportion of users who actually tried but did not manage to contribute remains to be specified, in order to determine if this button needs to be made more visible (e.g. by changing the color).

Transparency and traceability

Respondents were asked if they knew the existence of the procedure for the production and update of the Orphanet nomenclature, which details the quality standards applied to scientific data provided by Orphanet on rare diseases. 66% of them (N = 112) were unaware of the existence of is document.

It would be useful in future surveys to assess the proportion of users who are interested in reading this procedure for the production and update of the Orphanet nomenclature and, depending on the result, consider enhancing its visibility on the Orphanet website.

Perceived value

To assess qualities of the Orphanet nomenclature, users were asked what words they would use to describe it. 111 respondents selected one or more adjectives that they thought best described the Orphanet nomenclature among the eight following adjectives: Reliable, Up to date, Useful, Accurate (positive terms), Unreliable, Out of date, Not useful, Inaccurate (negative terms). Respondents selected 227 adjectives in total, of which 94% were positive terms, the most prominent was "Useful" (selected by 66% of respondents).

Feedback

Clients were asked to give additional feedback on improvements that could be made to the Orphanet nomenclature. 42% of all respondents (N=49) provided additional feedback. This is summarized as follows:

- improving the general information associated with rare diseases (42%), by keeping it up-to-date and make it easier to understand;
- improving the website interface (14%) for better access to the information,
- 12% provided positive feedback, stating that the Orphanet website is clear, intuitive, accessible, and provides the information they need.

Conclusions and issues for future considerations

Users are globally satisfied with the Orphanet nomenclature and the information that it provides. No significant difference was identified between response languages. A few elements for reflection have arisen regarding the accessibility of information:

- It would be interesting to introduce more detailed questions on the nomenclature of diseases (main name, synonyms, keywords) in future surveys, as it is presently difficult to ascertain why some users did not find the diseases they were looking for: whether it is because these were truly absent from Orphanet, or if the diseases are present in the database but the nomenclature is incomplete or lacks pertinence.
- Orphanet is developing a collaborative effort with European Reference Networks for the update of the nomenclature. This will greatly improve the efficiency of the scientific validation of data on rare diseases, and therefore enhance the accuracy, timeliness and completeness of the Orphanet nomenclature in view of the current knowledge.

II. Detailed satisfaction survey for the Orphanet rare disease classification

A total of 76 users chose to give their opinion the Orphanet classification of rare diseases.

The most represented response language was Spanish (45%), followed by Italian (29%), English (25%) and French (13%). The majority of respondents identified as health professionals (47%), followed by researchers (25%), patients, patient family members, or patient organizations (17%), and education or communication professionals (14%).

User experience with the classifications on the Orphanet website

Availability

21% of respondents (N = 76) did not know that the classification of rare diseases can be consulted on the Orphanet website, in which case the survey ended here.

Purpose

Of users who completed the rest of the survey (N = 60), 57% consult the classifications in order to obtain general information on how a disease is classified, 22% on the medical and/or surgical specialty, and 15% for coding purposes.

Finding the information

90% of respondents (N = 60) that consult the classifications stated that they usually find the classification(s) of interest and the associated disease(s)/clinical group(s) without difficulty.

For the 10% of users who have difficulty finding the classifications, the following reasons were selected in roughly equal proportions:

- access paths to classifications are not visible enough,
- the main name, synonyms, and/or keywords associated with the disease(s)/clinical group(s) that they were interested in were incomplete or lacked pertinence,
- the disease(s) or clinical group(s) of interest did not seem to be present in Orphanet.

Among users who consult the classifications in order to obtain information on the medical and/or surgical specialty, 90% agreed that they could easily or very easily identify this information, while 6% indicated that they did not manage to find it.

Among users who consult the classifications for coding purposes, 78% agreed that they can easily or very easily identify the ORPHAnumbers that they are looking for, while 22% disagreed with this statement.

These results, while generally positive, still emphasize some difficulty in accessing and searching for classifications in a subgroup of patients. An improvement has already been made in this regard: the level of classification of each clinical entity is now indicated on its respective disease page since July 2019 (after the survey was conducted), with a direct link to the classifications, as well as in the search result list. Future surveys will help determine the impact of these new features on the users' experience accessing and finding information on the classifications of rare disease in Orphanet.

Perceived value

Of the way the classifications are displayed on the Orphanet website

56 respondents selected one or more adjectives among the following: Convenient, Efficient, Easy to understand (positive terms), inconvenient, inefficient, and difficult to understand (negative terms).

Respondents selected 96 adjectives in total, of which 91% were positive terms. Only 8 users selected negative terms, the most prominent being inconvenient and difficult to understand.

Of the classifications in general

56 respondents selected one or more adjectives among the following: Reliable, Up to date, Useful, and Accurate (positive terms), Unreliable, Out of date, Not useful, and Inaccurate (negative terms).

Respondents selected 121 adjectives in total, of which 97% were positive terms, and the most prominent being "Useful" (selected by 66% of respondents).

Feedback

21% of all respondents (N=16) provided additional feedback on the Orphanet classifications, which mainly pertained to the way disease-related information and resources are displayed (25%): for example, 2 users commented on expert resources (e.g. expert centers and research projects), raising the fact that whether a given resource is specific to a disease or a group of diseases is not explicit enough. Another user suggested to make the classification "exportable" (e.g. in an Excel file). Note that classification datasets are already available in XML format on the Orphadata platform.

Conclusions and issues for future considerations

Users are globally satisfied with the Orphanet classifications and the information that they provide. No significant differences were identified between response languages.

The main field for future improvement would be the way classifications are displayed on the website: provide a dynamic display of the classifications, with a better and more intuitive visualization of diseases that are classified in several classifications.

III. Detailed satisfaction survey for cross-referencing of the nomenclature with other reference databases in the Orphanet website

General information

A total of 46 users chose to give their opinion on the cross-referencing of rare diseases in Orphanet. The most represented response languages were English and Spanish (13 respondents respectively), followed by Italian (6 respondents) and French (5 respondents).

The majority of respondents identified as health professionals (23), followed by patients, patient family members, or patient organizations (10), students (8), and a tie between researchers, healthcare managers/policy makers, and education or communication professionals (7 respectively).

Purpose

A majority of users (80%) consult the cross-referencing in order to obtain general information on the disease(s) of interest from other reference databases, while 15% indicated that they use it for coding purposes and interoperability with other coding systems (e.g. ICD-10, OMIM).

This is an encouraging result, as the purpose of the cross-referencing performed by Orphanet is to create a common node between the main coding systems used in various healthcare and research structures, and bring together information provided by different terminologies and databases with a focus on rare diseases.

Finding the information

83% of respondents indicated that they usually find the alignment(s) that they are looking for without difficulty.

Of the 17% of respondents who have difficulty finding the alignment(s):
- 4 attributed this difficulty to not being able to use external references other than OMIM and ICD-10 when using the Orphanet search engine.
- Other reasons, selected in equal proportion, included the nomenclature of disease(s) being incomplete or lacking pertinence (main name, synonyms, keywords), the disorder(s) of interest being seemingly absent from the Orphanet nomenclature, or the lack of alignment for these diseases.

Perceived value

42 out of the 46 respondents answered these questions, which were intended to assess the perceived usefulness of the cross-referencing of the Orphanet nomenclature with other reference databases and terminologies, and the general opinion of users on this service.

Usefulness of the cross-referencing

Globally, the result was very positive as 79% of respondents qualified the cross-referencing (all references included) as useful, including 31% who qualified it as "Very useful".

When broken down by item, the results show that the cross-referencing perceived as the most useful are OMIM and the ICD-10, for which the terms "Useful" and "Very useful" were selected by a total of 93% and 86% of respondents, respectively.

Regarding the cross-referencing that is perceived as the least useful, the results show a tie between UMLS and MedDRA, both considered as not useful ("not useful" or "not useful at all") by 31% of respondents. This result was expected, as these terminologies are a lot less commonly used than OMIM and ICD-10.
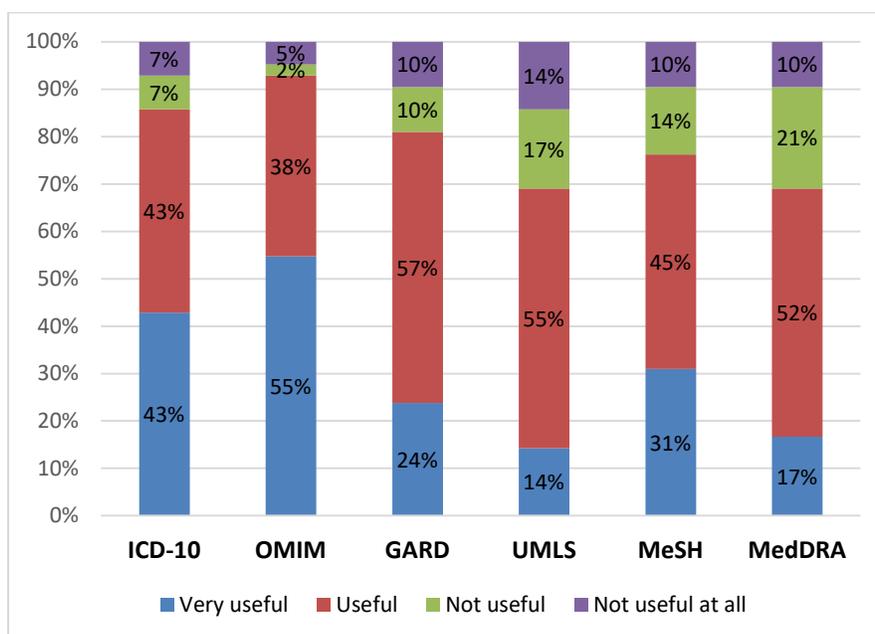
Figure 22: Perceived value of the Orphanet cross-referencing by database (n= 42). ICD-10 = International Classification of Diseases – 10th Edition; OMIM = Online Mendelian Inheritance in Man; GARD = Genetic and Rare Diseases Information Center; UMLS = Unified Medical Language System; MeSH = Medical Subject Headings; MedDRA = Medical Dictionary for Regulatory Activities

General perceived value of the cross-referencing

Respondents were asked to select one or more adjectives that they thought best described the cross-referencing of the Orphanet nomenclature among the following: Convenient, Easy to understand, Reliable, Up-to-date, Accurate (positive terms), Inconvenient, Difficult to understand, Unreliable, Out of date, Inaccurate (negative terms).

Respondents selected 99 adjectives in total, of which 91% were positive, the most prominent being "Convenient" (selected by 52% of respondents).

Transparency and traceability

78% of respondents (n= 41) were unaware of the procedure established by Orphanet for the attribution of ICD-10 codes for rare diseases that are not listed in the ICD.

Future surveys could help assess the proportion of users who are actually interested in reading this procedure and, depending on the result, consider enhancing its visibility on the Orphanet website.

Information redundancy

A majority of respondents (79%, n = 42) did not express any need for other cross-referencing than those already provided by Orphanet. Among the 21% remaining respondents, 56% cited other databases that they wished the Orphanet were cross-referenced with: SNOMED (Systematized Nomenclature of Human and Veterinary Medicine), HPO (Human Phenotype Ontology), PubMed, EMMeT (Elsevier Merged Medical Taxonomy), and Isabel Healthcare, a diagnostic tool used by clinicians to match clinical features with diseases.

Phenotypic features of rare diseases included in the Orphanet nomenclature are already annotated with HPO terms; and dedicated PubMed queries are available on the disease pages of a large number of clinical entities. Furthermore, a collaboration is ongoing between Orphanet and the International Health Terminology Standards Development Organization (IHT-SDO) to include rare diseases lacking in SNOMED-CT, and to provide alignments between ORPHAnumbers and SNOMED-CT terms.

Given their very small number, it may be useful to find out if the suggestions made by users to add cross-referencing and annotations that are already provided by Orphanet result from simple oversights on the users' part, or if they reflect a not-so-uncommon lack of awareness regarding the existence of these services, in which case the communication efforts around this subject would need to be improved.

Feedback

13 users (28% of all respondents) provided additional feedback on the Orphanet cross-referencing, among which 5 included suggestions for improvement. One user commented on the translation of cross-referencing into French, 2 on keeping the cross-referencing up-to-date, 1 on the accuracy, and one suggested accompanying cross-referencing with hypertext links on the disease page.

Conclusions and issues for future considerations

Users are globally satisfied with the cross-referencing of the Orphanet nomenclature. No significant differences were identified between response languages.

The perception of the OMIM and ICD-10 cross-referencing as the most useful is not surprising, since these coding systems are the most universally and globally recognized across healthcare and research systems. This further underlines the importance of the common node provided by ORPHAnumbers, which enable to discern and track information that specifically pertains to rare diseases among all diseases.

IV. Detailed satisfaction survey for the specialist clinics/ centres of expertise in the Orphanet website

A series of 6 questions were posed to users of the specialist clinics/ centres of expertise with the aim of assessing the usefulness of the information provided, its completeness and exhaustivity, as well as its clarity and accessibility (easy to find).

124 users replied to the questions concerning the specialist clinics/ centres of expertise. The majority of users described themselves as Patient/Family/Patient Organisation (33%) followed by the category of Health Professional (32%), of and researcher (11%).

Intended usage

This question aimed to assess the purpose(s) for which the directory of specialist clinics is used and thereby determine whether it is serving the needs of its users. Users could select multiple purposes with a minimum of at least one response. 96 respondents answered and selected 171 purposes.

Results show that users search for all the centres specialised in one or several diseases (38%) and contact details of the centre (34%). These results are surprising as Orphanet directory of services has been design in order to help users to identify the centre that is more convenient for them and not to have a landscape of all the centre available. These results suggest us that users might not have understood the question or possible responses. Next year question should be more precise in order to be able to draw conclusions.

User experience of specialist clinics /centres of expertise directory

Users were asked to evaluate the following qualities of the information provided by this directory. 94 users answered. They found the information was useful (84% agreed or strongly agreed), easy to read and understand (83%), and easy to find (79%).

Information redundancy

One question was dedicated on the usefulness of specific fields that are currently displayed on the website. This aimed at deciphering if the dataset shown could be simplified. For the majority of respondents (N=96), most of the fields were considered useful (>70%). However, the field for the fax number of the specialist clinics was highlighted as somewhat less useful with only 40% of the respondents considering it useful.

The inverse question was asked (what information is missing) in order to decide if more data should be collected by Orphanet. 50 respondents answered this question. 28% of the respondent answered that nothing was missing, 38% left comments showing that the current interface is not friendly enough as the extra information requested is already present on the website, 12% were interested in having more centre specific details (e.g. how the centre is functioning with the appointment scheduled, the delay to have an appointment). This kind of information is very difficult to maintain and update and might be found on the website of the clinic concerned; instead of this, Orphanet provides the clinic link where available. The remaining comments were on the coverage of the country covered by Orphanet, the need of a new/better search option or updates.

Feedback

The above result is concordant with the responses obtained to the question "Concerning the expert centres what could Orphanet do to better serve your needs?". Of the 48 persons that answered to this question, 23% answered that they were satisfied with this directory as it is, 21% of the responses showed that the interface was not friendly enough as people have difficulties to find the information they are looking for.

Perceived value

Users were asked the likelihood (from 0-10) they would recommend Orphanet directory of expert centres to a friend or colleague. 49 respondents answered 9 or 10, 28 answered 7 or 8 and 15 responded 6 or below, giving a NPS of 36.95.

Conclusion

Orphanet is working to improve the data update frequency, especially contact details. For search options, it is now possible to search by expert center. We are also implementing the "official designation" for genetic counseling that will be separated from the "official designation" of the medical management in order to increase transparency on the effective designations. A new type of network have been created called "Network of experts" to provide information on rare diseases networks, focused on either clinical expertise or research.

V. Detailed satisfaction survey for the patient organisation in the Orphanet website

A series of 6 questions were posed to users of the Orphanet patient organisations directory with the aim of assessing the usefulness of the information provided, its completeness and exhaustivity, as well as its clarity and accessibility (easy to find).

99 users replied to the questions concerning the patient organisation. The majority of users described themselves as Patient, family or related to patient Organisation (39%) followed by the category of health professional (22%), of and researcher (13%).

Intended usage

This question aimed at assessing whether the directory of patient organisations is serving the needs of its users. Users could select has many answers as they wanted with a minimum of one answer required. 74 respondents answered and selected 140 purposes.

Results show that users search equally for a specific patient organisation (34%), the contact details of the association (32%) or all the patient organisations in one or several diseases (30%). These results are surprising as Orphanet directory of services has been design in order to help users to identify the patient organisation that is more convenient for them and not to have a landscape of all the organisation available. These results suggest us that users might not have understood the question or possible responses. Next year question should be more precise in order to be able to draw conclusions.

User experience of patient organisations directory

Users were asked to evaluate the following qualities of the information provided by this directory. 73 users answered. They found the information was useful (73% agreed or strongly agreed), easy to read and understand (86%), and easy to find (60%).

Information redundancy

One question was dedicated on the usefulness of specific fields that are currently displayed on the website. This aimed at deciphering if the dataset shown could be simplified. For the majority of respondents (N=74), most of the fields were considered useful (>80%) However, the field for the fax number of the patient organisation was highlights as less useful, with only 20% of the respondents considering it useful.

The inverse question was asked (what information is missing) in order to decide if more data should be collected by Orphanet. 31 respondents answered to this question. 10 respondents answered that nothing was missing. 5 asked for a deeper coverage with the addition of patient organisations in countries not yet covered by Orphanet or by the addition of the various contact points in the same organisation. 4 were interested in having more details on what exactly the organisation is doing in term of services or activity. This kind of information is very difficult to maintain and update and might be found on the website of the concerned organisation; instead of this, Orphanet provides a link to the patient organisation where available. It is noteworthy that Orphanet collects only patient organisations that are non-profit organisations or foundations and which provide support and/or information to patients suffering from a rare disease. 3 responses were on the quality on the diseases linked to patient organisation as users find them not precise enough. 3 respondents also asked to have more updated patient organisation details. The remaining comments were disparate and concerned many different topics.

Feedback

The above result is concordant with the responses obtained to the question "Concerning the patient organisation what could Orphanet do to better serve your needs?". Indeed on the 29 persons that answered to this question, 24% answered that they were satisfied with this directory as it is, 7 % asked to have more updated data while 10% mentioned the coverage of the Orphanet directory of patient organisation.

Perceived value

Users were asked the likelihood (from 0-10) that they would recommend the Orphanet directory of patient organisations to a friend or colleague. 37 respondents answered 9 or 10, 19 answered 7 or 8 and 14 responded 6 or below, giving a NPS of 32.9.

Conclusion

Orphanet is working to improve the data update frequency, especially contact details. In order to direct users toward information concerning the activity of the patient organization, we will improve the visual presentation of the website links to the different associations. For search options, it is now possible to search by patient organization name.

VI.   Detailed satisfaction survey for the diagnostic tests in the Orphanet website

A series of 6 questions were posed to users of the Orphanet diagnostic tests and corresponding medical laboratory directory with the aim of assessing the usefulness of the information provided, its completeness and exhaustivity, as well as its clarity and accessibility (easy to find).

101 users replied to the questions concerning the diagnostic test. The majority of users described themselves as health professional (36%) followed by the category of patient, family or related to patient Organisation (21%), of and researcher (14%).

Intended usage

This question aimed at assessing whether the directory of diagnostic tests is serving the needs of its users. Users could select multiple answers with a minimum of one answer required. 68 respondents answered and select 116 types of usage.

The majority of users search for all the tests allowing diagnosis of one or several rare diseases (49%), this is followed by medical laboratory performing a specific diagnostic test (28%). These results are surprising as Orphanet directory of services has been design in order to help users to identify the diagnostic test (and its medical laboratory) that is more convenient for them and not to have a landscape of all the tests available. These results suggest us that users might not have understood the question or possible responses. Next year question should be more precise in order to be able to draw conclusions.

User experience of diagnostic tests directory

Users were asked to evaluate the following qualities of the information provided by this directory: usefulness, easy to find, read and understand. 67 users responded, of which the majority agreed or strongly agreed that the information was useful (91%), easy to read and understand (87%), and easy to find (87%).

Information redundancy

One question was dedicated on the usefulness of specific fields that are currently displayed on the website. This aimed at deciphering if the dataset shown could be simplified. For the majority of respondents (N=68), the majority of fields were considered useful (78%); however, two fields were highlighted as somewhat less useful and include i) the name of the director of laboratory and ii) the names of the person(s) responsible for the test, and were respectively considered useful for 50% and 62% of respondents.

The inverse question was asked (what information is missing) in order to decide if more data should be collected by Orphanet. 33 respondents answered this question. 13 respondents answered that nothing was missing. 15 asked us to add more details on the tests such as price, the methodology used and the test limitations. This kind of information is very difficult and time consuming to collect and maintain. Moreover, certain information is sensitive (such as cost) and may impact the willingness of the diagnostic labs to provide information, and thus may negatively impact repertory completeness. The remaining comments were disparate and concerned different topics.

Feedback

The above result is concordant with the responses obtained to the question "Concerning the diagnostic tests what could Orphanet do to better serve your needs?". Indeed, of the 27 persons that answered to this question, 26% answered that they were satisfied with this directory as it is. On the other hand, 37 % of the respondents requested more updated data which indicates that this directory could be more up-to-date.

Perceived value

Users were asked the likelihood (from 0-10) that they would recommend Orphanet directory of diagnostic tests to a friend or colleague. 30 respondents answered 9 or 10, 19 answered 7 or 8 and 14 responded 6 or below, giving a NPS of 25.4.

Conclusion

Orphanet is working to improve the data update frequency. Moreover, thanks to the feedbacks of professionals that use our online registration tools or that consult Orphanet, we are thinking of a new way to present our data in a more user friendly manner.

VII.   Detailed satisfaction survey for the registries in the Orphanet website

A series of 6 questions were posed to users of the Orphanet registries with the aim of assessing the usefulness of the information provided, the completeness and exhaustivity, as well as the clarity and accessibility (easy to find).

38 users responded to the questions concerning the registries. The majority of users described themselves as health professional (29%) followed by the category of patient, family or related to patient Organisation (21%), of and researcher (14%) and health care manger or policy maker (13%).

Intended usage

This question aimed at assessing whether the directory of registries is serving the needs of its users. Users could select multiple answers with a minimum of one answer required. 25 respondents answered and select 48 purposes.

Results show that users search equally for all the registries/ biobanks specialised in one or several rare diseases (36%), the address of specific registry/ biobank (33%) or the contact details of the manager of a specific registry/ biobank (31%). These results are surprising as Orphanet directory of services has been design in order to help users to identify the registry or biobank that is more convenient for them and not to have a landscape of all the registries or biobanks available. These results suggest us that users might not have understood the question or possible responses. Next year question should be more precise in order to be able to draw conclusions.

User experience of registries directory

Users were asked to evaluate the following qualities of the information provided by the directory: usefulness, easy to find, read and understand. Of the 67 respondents the majority agreed or strongly agreed that the information was useful (72%), easy to read and understand (75%), and easy to find (79%).

Information redundancy

One question was dedicated on the usefulness of specific fields that are currently displayed on the website. This aimed at deciphering if the dataset shown could be simplified. For the majority of respondents (N=25), most of the fields were considered useful (>70%). However, the field for the contact details of the funding body of the registry / biobank was highlighted as somewhat less useful with only 60% of the respondents considering it useful.

The inverse question was asked (what information is missing) in order to decide if more data should be collected by Orphanet. Of the 9 respondents, 3 indicated that nothing was missing, 3 requested more details on the registries and 2 for more updates.

Feedback

The above result is concordant with the responses obtained to the question "Concerning the registries what could Orphanet do to better serve your needs?". Indeed, of the 6 persons that answered to this question, 5 requested more updated data which indicates that this directory could be more up-to-date.

Perceived value

Users were asked the likelihood (from 0-10) they would recommend Orphanet directory of registries to a friend or colleague. 12 respondents answered 9 or 10, 8 answered 7 or 8 and 5 responded 6 or below, giving a NPS of 28.

Conclusion

Orphanet is working to improve the data update frequency. In order to give more information about the registry (if information is available), we will improve the visual of the page at the level of protocol links. For search options, it is now possible to search by registry name.

VIII. Detailed satisfaction survey for the research project in the Orphanet website

A series of 6 questions were posed to users of the Orphanet research projects with the aim of assessing the usefulness of the information provided, the completeness and exhaustivity, as well as the clarity and accessibility (easy to find).

90 users replied to the questions concerning the research projects. The majority of users described themselves as Patient, family or related to patient Organisation (28%) followed by the category of health professional (25%), of and researcher (20%).

Intended usage

This question aimed at assessing whether the directory of patient organisations is serving the needs of its users. Users could select multiple answers with a minimum of one answer required. 57 respondents answered and select 117 purposes.

Results show that users search in majority for all the research projects specialised in one or several rare diseases (42%) then for the contact details of the principal investigator of a specific research project (28%) and finally for where a specific research project is taking place (24%). These results are surprising and suggest us that they might not have understood the question or the different answer. Next year question should be more precise in order to be able to draw conclusions. Even so, these results show that the contact details of the principal investigator of a specific research project seems to be an important data for our users. This should be taking into consideration, as Orphanet tends to not collect this data anymore.

User experience of the research projects directory

Users were asked to evaluate the following qualities of the information provide by the directory: usefulness, easy to find, read and understand. 56 users answered. They found the information was useful (95% agreed or strongly agreed), easy to read and understand (93%), and easy to find (88%).

Information redundancy

One question was dedicated on the usefulness of specific fields that are currently displayed on the website. This aimed at deciphering if the dataset shown could be simplified. For the majority of respondents (N=57), most of the fields were considered useful (79%). However, the field for the funding body of the research project was highlighted as somewhat less useful with only 61% of the respondents considering it useful. These results show, as in the question in intended usage, that contact details of the investigator of the research project is useful for them. Thus Orphanet should pay special attention on this data.

The opposite question was asked (what information is missing) in order to decide if more data should be collected by Orphanet. Of the 21 respondents, 8 indicated that nothing was missing, 5 requested more details on the research project, specifically to include project description. As Orphanet systematically collects this description of the project, this evolution could be implemented and will be taken into consideration. The remaining comments were disparate and concerned different topics such as the inclusion of more countries or to have more updated data.

Feedback

The above result is concordant with the responses obtained to the question "Concerning the registries what could Orphanet do to better serve your needs?". Indeed, answers were in phase with the previous question, showing that Orphanet should pay special attention to the coverage and the update of its directory or to add more details on each research project.

Perceived value

Users were asked the likelihood (from 0-10) they would recommend Orphanet research projects directory to a friend or colleague. 29 respondents answered 9 or 10, 14 answered 7 or 8 and 12 responded 6 or below, giving a NPS of 30.9.

Conclusion

Orphanet is working to improve the data update frequency. In order to give more information about the project (where available), we will improve the visual of the page at the level of protocol links. For search options, it is now possible to search by research project name. A new type of network has been created called "Network of experts" to provide information on rare diseases networks focused either on clinical expertise or research.

IX. Detailed satisfaction survey for the clinical trial in the Orphanet website

A series of 6 questions were posed to users of the Orphanet clinical trial directory with the aim of assessing the usefulness of the information provided, the completeness and exhaustivity, as well as the clarity and accessibility (easy to find).

87 users replied to the questions concerning the clinical trials. The majority of users described themselves as Patient, family or related to patient Organisation (30%) followed by the category of health professional (26%), of and researcher (17%).

Intended usage

This question aimed at assessing whether the directory of clinical trials is serving the needs of its users. Users could select multiple answers with a minimum of one answer required. 56 respondents answered and select 150 purposes. Results show that users search equally for the clinical trials specialised in one or several rare diseases (28%), a specific clinical trial of interest (26%) or the location of a specific clinical trial (24%). These results are surprising as Orphanet directory of services has been design in order to help users to identify clinical trial focused on the disease they are interested in and not to have a landscape of all the trials on all rare diseases. These results suggest us that users might not have understood the question or possible responses. Next year question should be more precise in order to be able to draw conclusions. Even so, these results show that the location of the clinical trial seem to be an important data for our users. This should be taking into consideration, as Orphanet tends to not collect this data anymore.

User experience of the directory of clinical trial

Users were asked to evaluate the following qualities of the information provided by this directory: usefulness, easy to find, read and understand. Of the 55respondents, the majority agreed or strongly agreed that the information was useful (89%), easy to read and understand (82%), and easy to find (85%).

Information redundancy

One question was dedicated on the usefulness of specific fields that are currently displayed on the website. This aimed at deciphering if the dataset shown could be simplified. For the majority of respondents (N=56), most of the fields were considered useful (>84%). However, two fields were highlighted as somewhat less useful and include i) the Contact details of the sponsor of the clinical trial and ii) the contact details of the funding body(ies) of the clinical trial, for which only 53% and 62% of the respondents found useful respectively.

The inverse question was asked (what information is missing) in order to decide if more data should be collected by Orphanet. Of the 17 respondents, 10 indicated that nothing was missing, 2 requested more updated data and 2 more details. The remaining comments were disparate and concerned many different topics.

Feedback

The above result is concordant with the responses obtained to the question "Concerning the clinical trial what could Orphanet do to better serve your needs?". Indeed, of the 15 persons that answered to this question, 47% answered that they were satisfied with this directory, 20 % asked to have more details on clinical trials while 7% desired more updated data.

Perceived value

Users were asked the likelihood (from 0-10) they would recommend Orphanet disease clinical trial directory to a friend or colleague. 23 respondents answered 9 or 10, 21 answered 7 or 8 and 9 responded 6 or below, giving a NPS of 26.4.

Conclusion

Orphanet is working to improve the data update frequency. In order to give more information about the clinical trial (where available), we will improve the visual of the page at the level of protocol links. For search options, it is now possible to search by clinical trial name.

X. Detailed satisfaction survey for the Orphan Drugs in the Orphanet website

A series of 6 questions were posed to users of the Orphanet Orphan drugs directory with the aim of assessing the usefulness of the information provided, the completeness and exhaustivity, as well as the clarity and accessibility (easy to find).

69 users replied to the questions concerning the clinical trials. The majority of users described themselves as health professional (36%) followed by the category of patient, family or related to patient Organisation (23%), of and researcher (14%).

Intended usage

This question aimed at assessing whether the directory of clinical trials is serving the needs of its users. Users could select multiple answers with a minimum of one answer required. 40 respondents answered and select 75 purposes.

Results show that users search almost equally for all drugs concerning one or several rare diseases (39%), a specific drug (33%) or the stage of development of a specific drug (27%). These results are surprising as Orphanet directory of services has been design in order to help users to identify the drugs intended for the disease they are interested in and not to have a landscape of all the drugs available for all the diseases. These results suggest us that users might not have understood the question or possible responses. Next year question should be more precise in order to be able to draw conclusions.

User experience of the directory of Orphan drugs

Users were asked to evaluate the following qualities of the information provided by this directory: usefulness, easy to find, read and understand. Of the 40 respondents, the majority agreed or strongly agreed that the information was useful (90%), easy to read and understand (85%), and easy to find (83%).

Information redundancy

One question was dedicated on the usefulness of specific fields that are currently displayed on the website. This aimed at deciphering if the dataset shown could be simplified. For the majority of respondents (N=40), most of the fields were considered useful (>75%). However, the field for the code name of the Orphan drug was highlighted as somewhat less useful, with only 65% of the respondents considering it useful.

The inverse question was asked (what information is missing) in order to decide if more data should be collected by Orphanet. Of the 11 respondents, 5 indicated that nothing was missing, 2 requested information on the side effects of the drugs, and 2 requested the different undisplayed details. The remaining comments were about the stage of development (1) and the update that should be more frequent (1).

Feedback

The above result is concordant with the responses obtained to the question "Concerning the Orphan drugs what could Orphanet do to better serve your needs?". Indeed, of the 8 persons respondents, 3 answered that they were satisfied with this directory, the remaining comments were disparate and include more updated data, more details for each orphan drugs, to indicate their prices.

Perceived value

Users were asked the likelihood (from 0-10) they would recommend Orphanet disease clinical trial directory to a friend or colleague. 19 respondents answered 9 or 10, 10 answered 7 or 8 and 9 responded 6 or below, giving a NPS of 26.3.

Conclusion

Orphanet is working to improve the data update frequency.

XI. Detailed satisfaction survey for the Diseases summary texts

A series of 6 questions were posed to users of the disease summary texts with the aim of assessing the pertinence of the information provided, the completeness and exhaustivity of the texts, as well as the clarity and accessibility (findability) of the texts.

In this section, 114 users replied to these questions, the majority of which described themselves as a Health Professional (60%), followed by the category of Patient/Family/Patient Organisation (29%), and Research (19%).

Intended usage

This question aimed to assess whether the Orphanet disease summary text is serving the needs of its users or identify if there is redundancy in the texts. Users could select multiple answers with a minimum of one answer required. 114 respondents answered.

Results show that the user typically searches for general information on a disease, usually with a view to finding information on diagnosis, treatment and prognosis, and genetic counselling. Users did not tend to look for a specific section only, with many users selecting multiple categories. This suggests that all sections are useful to our users and that there is little redundancy.

User experience of the disease summary texts

This question (114 responses) aimed at assessing the following qualities: clarity of the texts, whether the information is informative, of good quality and how easy they are to find. The majority of users agreed or strongly agreed that the texts are clear (94%), informative (94%), of good quality (96%) and easy to find (92%).

To assess other qualities of the texts, users were asked what words they would use to describe them (obligatory question, 114 responses). The most frequent descriptions included useful (73% of responses), reliable (57%, and concise (49%). However, areas identified for improvement include:

- the amount of information/details provided in the text (with 30% of responses describing the texts as either incomplete or contain too little information)
- the frequency of updates (9% of responses)

Perceived value

To assess the overall satisfaction with the encyclopaedia and its perceived value, users were asked to rate their satisfaction (very satisfied, somewhat satisfied, somewhat dissatisfied). Overall satisfaction of the diseases summary texts is high, with 49% very satisfied, 44% somewhat satisfied, and only 7% of users were somewhat dissatisfied with the texts. For the dissatisfied users, no significant tendency was found (e.g. specific type of user or looking for specific information).

Users were also asked the likelihood (from 0-10) that they would recommend Orphanet disease summary texts to a friend or colleague. 66 respondents answered 9 or 10, 33 answered 7 or 8 and 12 responded 6 or below, giving a NPS of 49.

Feedback

Clients were asked to give additional feedback on improvements that could be made to the diseases summary texts for which 43 out of 114 respondents provided feedback, which is summarised below:

- **Text detail**: 42% of respondents (N=17) indicated that generally more detail is required in the texts, with certain sections/themes were highlighted such as the functional consequences or the details on treatment and medication.

This result is in concordance with the one of the first part of the survey where 100 respondents suggested that the abstracts require more detail in general or on particular sections.

- **Provide up to date texts**: 19% of respondents (N=8) indicated texts update frequency was the next major area of improvement. In particular, users highlighted that many texts are over a decade old.

This result is in concordance with the one of the first part of the survey, where approximately 150 users commented on frequency of updates, and is an issue currently being addressed (as detailed in section **Erreur ! Source du renvoi introuvable.**).

- **Completeness of database:** 12% of respondents (N=5) raised the fact that currently many diseases do not have texts or definition. This problem should diminish with time as many texts are currently under validation
- **Intelligibility of the texts**: 12% of respondents (N=5) raised text comprehension; however, these users were predominantly searching in the capacity of patient, family or patient organization. Suggestions to improve accessibility to non- medical professionals include inclusion of a medical dictionary, where complicated terminology is linked to the definition or explain more clearly for non- patients

Conclusions on the user's satisfaction on the disease summary texts

Overall the satisfaction is very high for the disease summary texts; however, the results of the survey have highlighted three areas which the Encyclopedia for professionals should prioritize:

- increase the amount of detail in abstracts,
- increase the number of diseases with abstracts
- reduce the number of out of date texts (currently in progress)

XII.   Detailed satisfaction survey for epidemiological data in the Orphanet website

In this section, 45 users replied to the questions concerning the collection of epidemiological data in the Orphanet website. French speakers were the largest contributors to the questionnaire with 28.9% of all respondents. Italian (26.7%), Spanish (26.7%) and English (13.3%) account for most of the remaining respondents.

The respondents were primarily health professionals (36%), with researcher making 19% of respondents and patients, family members or patient organisations 14%.

Question 1: How often do you consult Orphanet to obtain epidemiological data on rare disease? N=45

To determine visiting frequency, users were asked how frequently they consult Orphanet in order to obtain epidemiological data. N= 45, only one choice was possible.

In general, the respondents consult Orphanet for epidemiological data on a regular basis, with 42% of users visiting once a month, and 22% once a week. The rest of the respondents consult epidemiological data in Orphanet once a year (25%) or less (11%)

Question 2: To what extent do you agree or disagree with the following statement? N=45

This question aims to evaluate the usability and comprehensiveness of epidemiological information. Only one response was possible
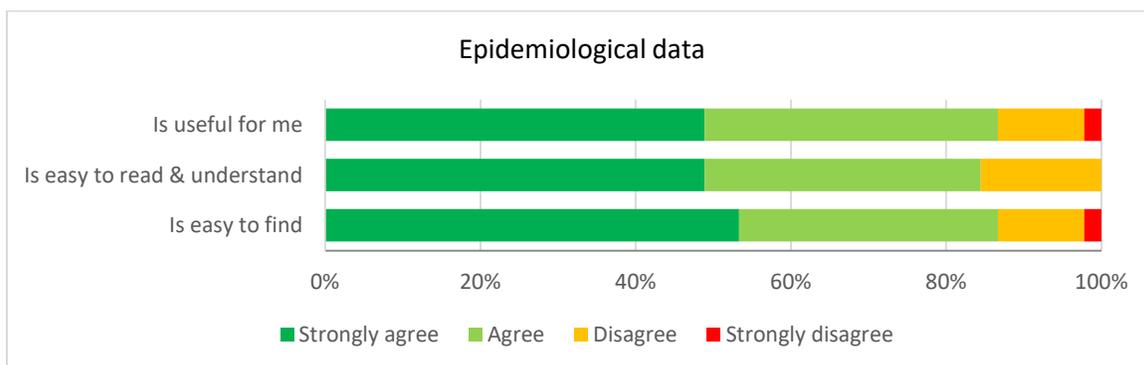

Figure 23: User experience of epidemiological data collected by Orphanet (N=45).

The vast majority of the respondents agreed or strongly agreed that the epidemiological data are useful, easy to find, and easy to read and understand. It should be, however, noted that to the question what Orphanet could do to better serve your needs, around 7% of the respondents asked that the epidemiological data should be presented more clearly.

Question 3: Overall, how satisfied are you with the Orphanet epidemiological data? N=45

The satisfaction of users was evaluated through this question. Most of the respondents were very satisfied (42%) or somewhat satisfied (45%). Those who were somewhat dissatisfied and very dissatisfied represent 11% and 2%, respectively.

Question 4: Concerning epidemiological data what could Orphanet do to better serve your needs? N=15

15 respondents, representing 33.3% of the persons that agreed to respond to this product, provided comments.

Suggestions mainly focused on the need to cover more geographical areas, to present the data more clearly, including the details, particularly with regard to the geographic area covered by the information provided. Moreover, the source of information used for annotations was one of the main requests made by the respondents.

Question 5: How likely is it that you would recommend Orphanet for its epidemiological data on rare disease to a friend or colleague?

This question aimed to ascertain how likely the respondent was to recommend Orphanet for its epidemiological data on rare diseases to a friend or colleague by asking them to give a score of 0 to 10 (1 being not at all likely, 10 being very likely).

The net promoter score was 31.2, which must be considered as a starting point in measuring our customer satisfaction that we will track over time.

Even if the sample of respondent is small for these products, indicative trends in the users' satisfaction have emerged, especially when we compare these results to the first part of the survey. Globally, users are happy with the epidemiological data that Orphanet provides but efforts should be made concerning display of the data as well as the associated references. It is noteworthy that one of the future informatics development will be to add the region concerned (i.e. worldwide or European) to the class of prevalence displayed on the disease page. Moreover, Orphanet is considering options for displaying the epidemiological data collected for each country. Concerning the source of the epidemiological data, these latter are accessible in the "Rare disease epidemiology" product that is freely accessible on Orphadata.

XIII. Detailed satisfaction survey for the natural history of the disease

In this section, 80 users replied to the questions concerning the collection and display of natural history of the disease (inheritance and age of onset of the disease).

Italian (32.5%) and Spanish (27.5 %) speakers were the largest contributors to the questionnaire. French (20%) and English (12.5%) account for most of the remaining respondents.

The respondents were primarily health professionals (41%), followed by patients, family members or patient organisations (22%) and then researchers (13%).

Question 1: How often do you consult Orphanet to obtain natural history information on rare disease? N = 80

This question aims to determine visiting frequency of respondents when searching for information on age of onset and inheritance. Only one choice was possible.

The majority of respondents consult Orphanet to obtain information on age at onset and the mode of inheritance once a month on a regular basis with 55% visiting once a month, and 24% visiting once a week. The rest of the respondents consult these data in Orphanet once a year (14%) or less (7%)

Question 2: To what extent do you agree or disagree with the following statement? N=80

This question aims to evaluate the usability and comprehensiveness of epidemiological information. Only one response was possible.
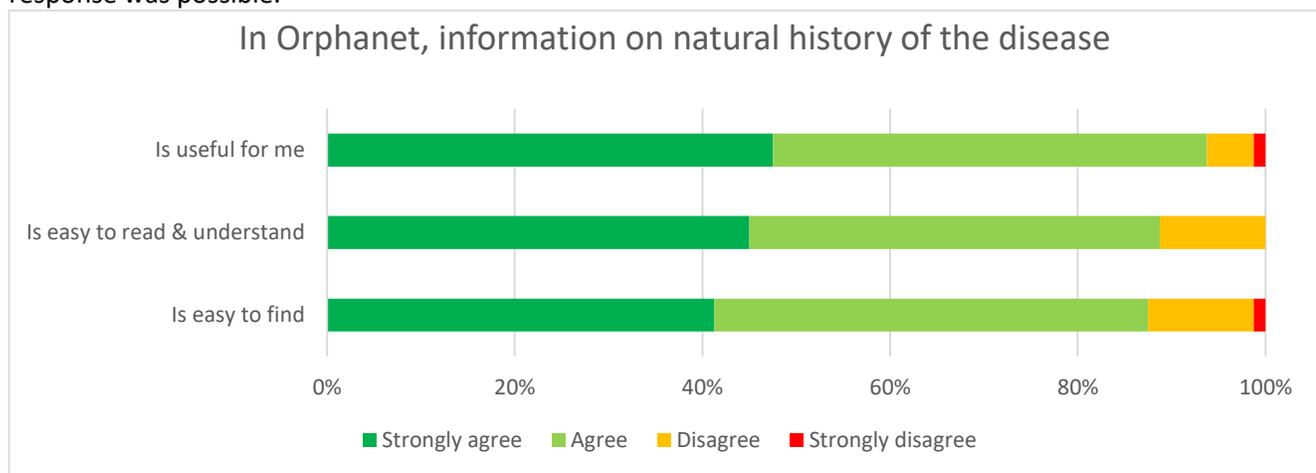

Figure 24: User experience of natural history data collected by Orphanet (N=45).

The vast majority of the respondents agree or strongly agree that information on natural history of the disease is useful (94%), easy to find (88%), read and understand (89%). However, it should be noted that in the question "what could Orphanet do to better serve your needs?", around 10% of the respondents asked that the data on natural history of the disease should be presented more clearly.

Question 3: Overall, how satisfied are you with the Orphanet natural history of the disease? N=80

The satisfaction of users was evaluated through this question. Most of the respondents were very satisfied (49%) or somewhat satisfied (41%). Those who were somewhat dissatisfied and very dissatisfied represent 9% and 1%, respectively.

Question 4: Concerning epidemiological what could Orphanet do to better serve your needs? N=24

24 respondents provided comments, representing 30% of the users that agreed to respond on this product. Suggestions mainly focused on the need to give more details on the natural history of the disease, to present the data more clearly, to update more frequently these data and to provide the sources of information used.

Question 5: How likely is it that you would recommend Orphanet for its natural history data on rare disease to a friend or colleague?

This question aimed to ascertain how likely the respondent was to recommend the natural history of the disease to a friend or colleague by asking them to give a score of 0 to 10 (1 being not at all likely, 10 being very likely).

The net promoter score was 33.8, which must be considered as a starting point in measuring our customer satisfaction that we will track over time.

Even if the sample of respondent is small for this product, indicative trends in the users' satisfaction have emerged, especially when we compare these results to the first part of the survey. Globally, respondents are happy with natural history of the disease provided by Orphanet but efforts should be made concerning their display and providing the associated references. We take the opportunity of this document to remind our user that an Orphadata product on natural history of rare disease exist and is freely available (http://www.orphadata.org/cgi-bin/epidemio.html)

XIV. Detailed satisfaction survey for the genetic information provided by Orphanet

49 users gave answers to the questions on gene information. Spanish speakers were the largest contributors to the questionnaire with 35% of all respondents. Italian (27%), English (18%) French (8%) and Portuguese (8%) account for almost all the remaining respondents.

The respondents were primarily health professionals (31%), followed by students (25%), researches (18%) and then patients, family members or patient organisations (15%).

Question 2: How often do you consult Orphanet to obtain information on gene(s) concerning a rare disease?

This question aims to determine visiting frequency of respondents when looking for information on gene(s). Only one choice was possible between « once a week » ; « once a month » ; « once a year » ; « less than once a year ».

The majority of respondents consult Orphanet to obtain information on gene on a regular basis with 47% consulting the website once a month and 31% once a week. The rest of the respondents consult these data in Orphanet once a year (16%) or less (6%)

These results emphasise the importance to have updated genes-disease relationship at all times. This is why we are currently working on the display of this information in the disease summary text. The idea behind this evolution is to extract from the database the genes-disease relationships of all the genes involved in a disease and to display this information directly in the disease summary text.

## Question 3: To what extent do you agree or disagree with the following statement? N=49
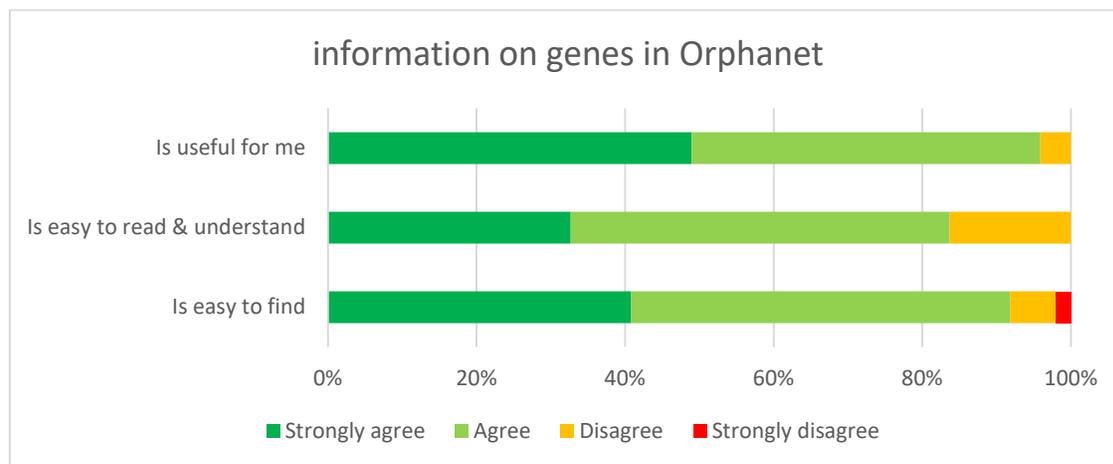
Figure 25: User experience of genetic information collected by Orphanet (N=49).

This question aims to evaluate the usability and comprehensiveness of gene-related information. Only one response was possible. The vast majority of the respondents agree or strongly agree that the genetic information is useful, easy to find, read and understand.

Question 4: According to your needs, which information displayed on the Orphanet website are unnecessary for the gene?

This question aimed to determine redundancy (if any) in gene-associated information displayed on the website (e.g. synonyms, OMIM, HGNC number, number of tests linked). Respondents could select multiple choices with no minimum requirement. None of the proposed items were selected by the majority of the respondents. The field considered the most unnecessary was the previous symbols and names of gene (selected by 35% of the

respondents). The low number of respondents and the lack of clear consensus does not allow us to make decision on this topic and should be further investigated further before simplify the gene page.

Question 5: According to you, which information are missing for the gene? And what could Orphanet do to better serve your needs?

The first part of this question aimed at detecting some missing information that the Orphanet users need. Respondents were invited to leave a comment (not obligatory). Of the 49 respondents, 88% either did not comment or indicated that nothing was missing. The second part of this question was design to detect some unfulfilled needs or services that Orphanet could consider to develop in order to increase its utility towards its users.

Most of the information the respondents judged as missing (contact to send new mutations/ inheritance/procedure details and information about how to live with genetic disease) are, in fact, present on Orphanet website. This results raises the fact that the website is not user-friendly enough and that information are not easy to find. Respondents also asked for links towards other databases but they do not specify which database(s). Lastly, one respondent suggested the need to improve the way in which gene information is presented. This development, however, is currently in progress.

Question 6: How likely is it that you would recommend Orphanet for its gene information on rare disease to a friend or colleague? N=49

This question aimed to ascertain how likely the respondent was to recommend Orphanet data to a friend or colleague by asking them to give a score of 0 to 10 (1 being not at all likely, 10 being very likely).

The net promoter score was 30 which must be considered as a starting point in measuring our customer satisfaction that we will track over time

XV. Detailed satisfaction survey for indexation of functional consequences of rare diseases

In this section, 45 users replied to the questions concerning the indexation and display of rare diseases with disabilities in Orphanet.

French speakers were the largest contributors to the questionnaire with 33% of all respondents. Italian (27%), Spanish (22%), English (9%) and German (7%) account for the remaining respondents.

The respondents were primarily patients, family members or patient organisations (40%), followed by health professionals (25 %) and then researchers (11%) and education/communication experts (11%).

Question 1: To what extent do you agree or disagree with the following statement? N=45

This question aims to evaluate the usability and comprehensiveness of the indexation of the functional consequences of the disease. Only one response was possible per question.
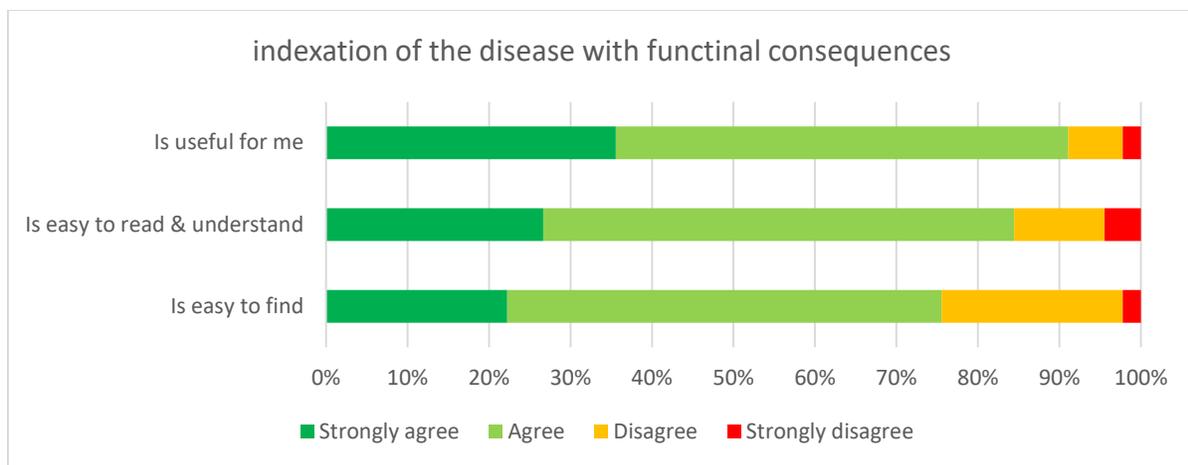

Figure 26: User experience of indexation of functional consequences of rare diseases collected by Orphanet (N=45).

The majority of respondents agreed or strongly agreed that the indexation of functional consequences is easy to find (75%), easy to read and understand (85%), and useful (91%). This is reassuring due to the complexity of presenting this information online in a way that is easily understood. A lesser, but significant number, of respondents disagreed or strongly disagreed and found the data difficult to find (25%) as well as difficult to read/understand (15%). This result is concordant with what respondent answered during the first phase of this survey when they asked to Orphanet to develop such indexation indicating that they did not know this product was already present in Orphanet. Despite evidence that most respondents are satisfied with these three criteria, it is clear that there is much room for improvement in the way that the functional consequences can be presented, which will ultimately increase usability and understanding.

Question 2: Which of the following words would you use to describe Orphanet annotations of the functional consequence of rare disease?

This question was aimed to ascertain in more detail what people think of the Orphanet data on functional consequences. Respondents could choose multiple adjectives in the list with no minimum requirement (not obligatory). 45 users responded to this question and selected a total of 122 adjectives. The largest number of responses was very positive adjectives: useful (24%), informative (21%) and reliable (21%).
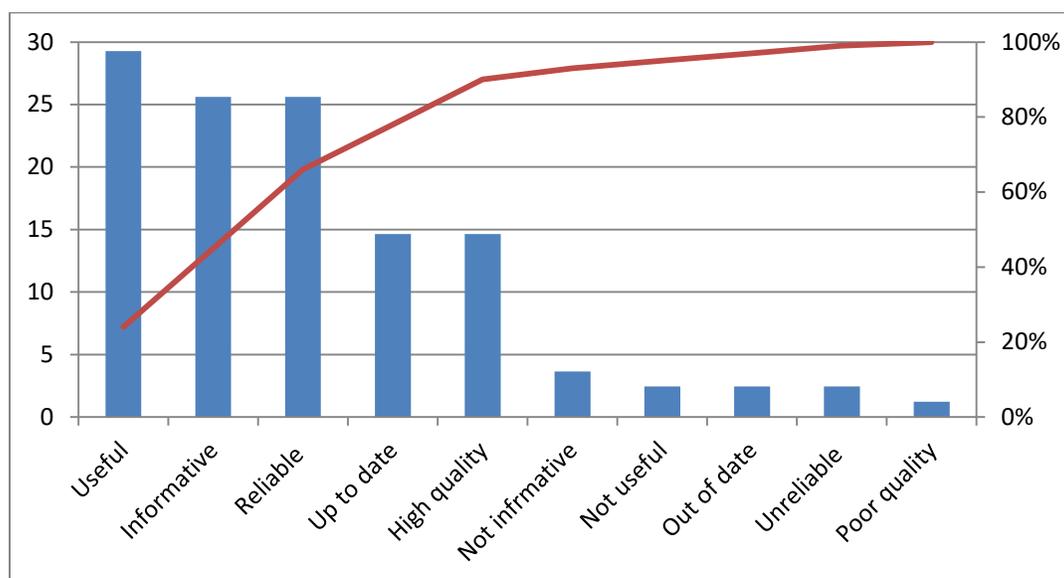
Figure 27: Pareto graphic representing which of the following words the respondents would use to describe Orphanet annotations of the functional consequence of rare disease

The number of negative responses (not useful, out of date, etc.) was limited and further reinforces the fact that the functional consequences indexations are highly appreciated by those that use them.

Question 3: How likely is it that you would recommend Orphanet data on functional consequences of rare diseases to a friend or colleague? N=45

As seen in previous questions, the majority (45%) of respondents would highly recommend the indexations of functional consequences produced by Orphanet. In addition, 36% of respondents gave a high score (8-7) while a much smaller number of respondents (20%) gave scores ranging from 6 to 0. This gave an NPS of 25. It is important to note that one respondent did not answer this question, but we interpreted the answer as a 0 based on their other responses throughout the questionnaire.

Question 4: Concerning the indexation of the functional consequences of rare diseases what could Orphanet do to better serve your needs? N=17

Open-ended comments were provided by 17 individuals in response to this question. Open-ended suggestions, as mentioned above, are important to the Disability project due the collaborative nature of the indexations as well as the factsheets. Most of those that did respond had no opinion or no specific suggestions.

Suggestions made were classified into three categories based on their general theme:

- improvement in terms of clarity and precisions. Although these suggestions are both short and vague ('Specify details' or 'Great clarity'), it is easy to imagine how additional notes and, most certainly, an improvement to the user interface would aid comprehension.

- The need to increase the coverage of diseases and syndromes. 9% of respondents pointed out the fact that the coverage of rare disease is still low. Orphanet should make clear that our immediate goal is to cover the most prevalent rare diseases. Also, the collaborative nature of this project makes the data production slow. A different respondent mentioned that an entire family of pathology could not be found, which reinforces this theme. The number of indexations must increase substantially as well improving the coverage of indexations amongst different pathologies.

XVI. Detailed satisfaction survey for the disabilities factsheets of rare diseases

A series of 5 questions were posed to users of the disabilities factsheets with the aim of assessing the clarity and accessibility (findability) of the texts

In this section, 51 users replied to these questions, the majority of which described themselves as a patient/family member/patient organization (49%), followed by Health professionals (46%). The rest of the respondents are fairly evenly split between research, health managers/policy makers, education/communication and students (11-14%). The majority of respondents were French (39%) followed by Italian (22%) and Spanish (19%) while these factsheets only exists in French

Intended usage

This question aimed to assess whether the Orphanet disabilities factsheet is serving the needs of its users. Users could select multiple answers within the following list: a description of the disease, the disability-related measure of the disease, the consequences in everyday life of the disease.

The majority (90%) use the disability factsheet to search for "The consequences in everyday life of the disease". Other information that is highly sought after (>75%) included "A description of the disease" and "The disability-related measure of the disease".

User experience of the disease summary texts

This question (51 responses) aimed at assessing the following qualities: clarity of the texts, whether these texts are useful for the reader and how easy they are to find.

The majority of respondents (90%) find the texts easy to read and understand. A small proportion of respondents (10%) do not find the texts easy to read and understand. Over half of clients who found the factsheets difficult to read and understand were patients/family/patient organisations.

The majority of the respondents find the texts useful, with 10% of clients finding the texts not useful. Over half of the clients who did not find the texts useful were patients/family/patient organisations. Further investigation is required to find out why 10% users do not find the disability fact sheets useful. The majority of clients (82%) describe the factsheets easy to find. 18% do not agree that the texts are easy to find. Two thirds of clients who find the texts difficult to find are patients/family/patient organizations. Restructuring of website is currently in the stages of development to improve user experience and findability of the different resources
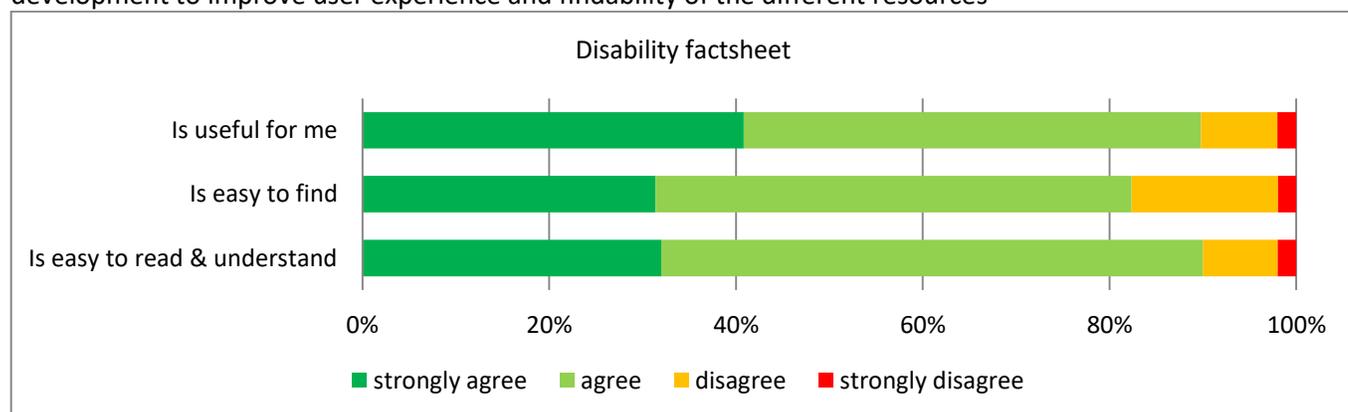

Figure 28: User experience of the disability factsheets produced by Orphanet (N=51).

To assess other qualities of these texts, users were asked what words they would use to describe them (114 responses). The most frequent descriptions included informative (21% of responses), reliable (18%), useful (18%) and up-to-date (13%). 10% of the responses where negative as split as follow:

- The quantity of information provided (described by 6 respondents as having both too little and by 4 respondents as having too much information (2%)
- The comprehensibility and the update of the texts (2 responses for both)
- The length and usefulness (1 response each)
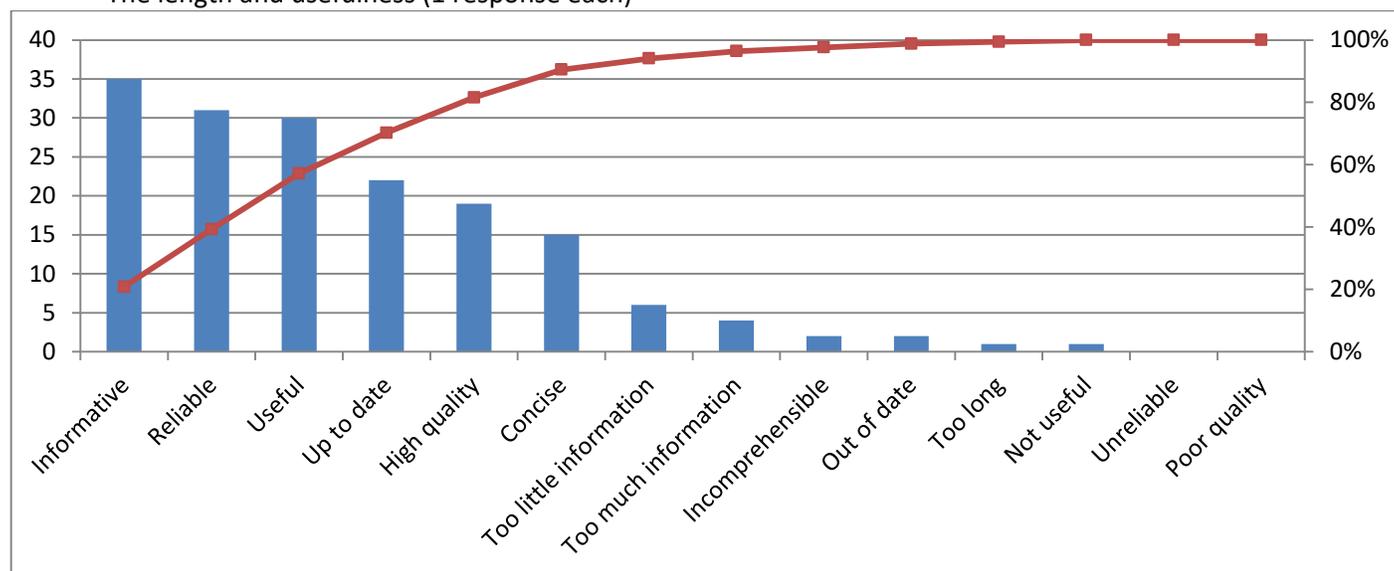
Figure 29: Pareto graphic representing which of the following words the respondents would use to describe Orphanet annotations of the functional consequence of rare disease

Perceived value
Users were asked the likelihood (from 0-10) that they would recommend Orphanet disability factsheets to a friend or colleague. 25 respondents answered 9 or 10, 16 answered 7 or 8 and 7 responded 6 or below, giving a NPS of 37.5.

Feedback
Clients were asked to give additional feedback on improvements that could be made to the disability factsheets for which 19 out of 51 respondents provided feedback. These latter concerned the content of the factsheets (32%), the number of disease covered (21%), the visibility/accessibility of the factsheets (16%) and the frequency of the updates (11%).

Conclusions on the user's satisfaction on the disease summary texts
Overall the satisfaction of the respondents for the disability factsheets tends to be high. Whilst this survey has given focus handicap some clues on how to improve, it is difficult to make any conclusions due to the low number of responses. Despite this three areas for consideration include:
- Increasing the visibility of the factsheets which relies on restructuring of website
- Increasing of the texts clarity for patients/family/patient organizations by
  o Exploring the possibility of including a table of content to better direct users, and help them bypass the more technical first page
  o Exploring the possibility of including a list of definitions or a medical dictionary. Avoiding clinical terminology is not considered as the texts are aimed also at health professionals, particularly in France where they are used for determining disability allowance and support.
  o Increase utility for the patients/family/patient organizations. This axis of improvement is worth asking users specifically how to increase utility or why not useful to them in orders to be as efficient as possible.

XVII. Detailed satisfaction survey for the emergency guidelines on rare diseases

A series of 7 questions were posed to users of the emergency guidelines with the aim of assessing the pertinence of the information provided, the completeness and exhaustivity of the texts, as well as the clarity and accessibility (findability) of the texts.

In this section, 39 users replied to these questions, the majority of which described themselves as a Health Professional (46%), followed by the category of Patient/Family/Patient Organisation (21%), and Education/communication (9%).

Intended usage

This question aimed to assess whether the Orphanet emergency guidelines are serving the needs of its users or identify if there is redundancy in the texts. Users could select multiple answers with a minimum of one answer required. 114 respondents answered.

Results show that the user almost equally searches for the different types of emergency situations and immediate therapeutic measures, the general emergency recommendations, the drug precautions. Users did not tend to look for a specific section only, with many users selecting multiple categories. This suggests that all sections are useful to our users and that there is little redundancy.

User experience of the disease summary texts

This question (39 responses) aimed at assessing the following qualities: clarity of the texts, and how easy they are to find. The majority of users agreed or strongly agreed that the texts are clear (94%) and easy to find (92%).

To assess other qualities of the texts, users were asked what words they would use to describe them (140 responses). The most frequent descriptions included reliable (16% of responses), informative (14%), and useful (14%). However, areas identified for improvement include:

- the amount of information/details provided in the text (with respectively 4%, 1% and 1% of responses describing the texts as containing too little information, being not informative and of poor quality)
- the format of the text described as being not practical With 3 % of the responses
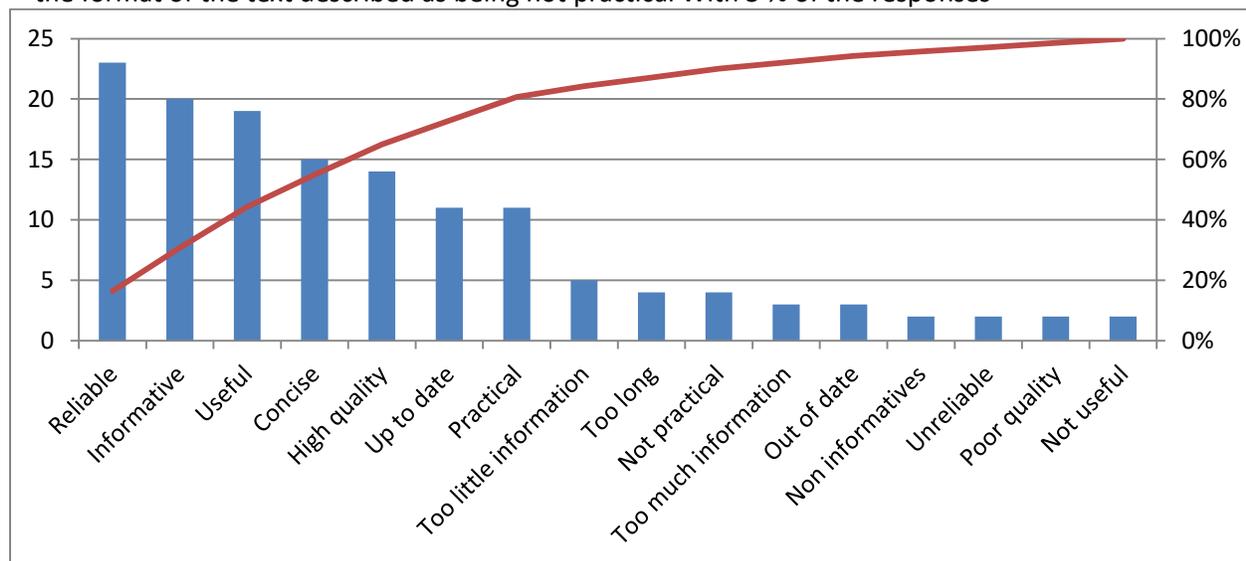

Figure 30: Pareto graphic representing which of the following words the respondents would use to describe Orphanet emergency guidelines

Perceived value

To assess the overall satisfaction with the emergency guidelines and its perceived value, users were asked to rate their satisfaction (very satisfied, somewhat satisfied, somewhat dissatisfied). Overall satisfaction of the diseases

summary texts is high, with 38% very satisfied, 56% somewhat satisfied, and only 5% of users were somewhat dissatisfied with the texts. For the dissatisfied users, no significant tendency was found (e.g. specific type of user or looking for specific information).

Users were also asked the likelihood (from 0-10) that they would recommend Orphanet disease summary texts to a friend or colleague. 16 respondents answered 9 or 10, 5 answered 7 or 8 and 3 responded 6 or below, giving a very good NPS of 54.

Feedback

Clients were asked to give additional feedback on improvements that could be made to the emergency guidelines for which 10 respondents gave feedback on areas of improvement for the emergency guidelines. Themes highlighted include:

- Increasing the disease coverage of the guidelines i.e. more diseases
- Increasing the text completeness or suggestions for structure (e.g. use diagrams and flowcharts for diagnostics and therapy, and guidelines organized by symptoms)
- Increase the accessibility or promotion of the guidelines
- Keeping the guidelines updated, minimum every 5 years

Conclusions on the user's satisfaction on the disease summary texts

Generally very positive response; however, not enough responses received to make firm conclusions on how to improve the guidelines. It would be useful to investigate further propositions made in feedback (e.g. schematics for work flow).

An additional survey is proposed targeted at urgentists at the FMSU conferences to get specific feedback on how to increase practicality of the emergency guidelines. In terms of understanding the need for promotion of the guidelines, suggestion to ask who amongst the urgentists have heard or used the emergency guidelines before.

D. RESULTS OF THE SATISFACTION SURVEY ON ORPHADATA (DATASETS FOR RESEARCH)

In this section, 38 users replied to the questions concerning the data download platform Orphadata. The respondents were primarily patients, family members or patient organisations (26%), followed by health professionals (22%) and researchers (17%). The result is surprising as this service is intended primarily for the research field.

How often do you download data from Orphadata? N =38:

This obligatory question aimed to find out the frequency at which users downloaded datasets from Orphadata. There were 38 replies, showing that just over half of the respondents (53%) downloaded data monthly. As most datasets are generated on a monthly basis, it was surprising to see that some users download data weekly: how to best sign post the update frequency will be considered.

Which datasets are you interested in/ have you already downloaded? N =38

Respondents were invited to indicate which datasets they were interested in (Figure 31 Interest in/ downloading of freely accessible or on request datasets) and which datasets they had already downloaded. Respondents were invited to select all options that applied to them, but it can be seen from the results that this was not well understood by the respondents. This, and the small sample size make interpretation of the results difficult.
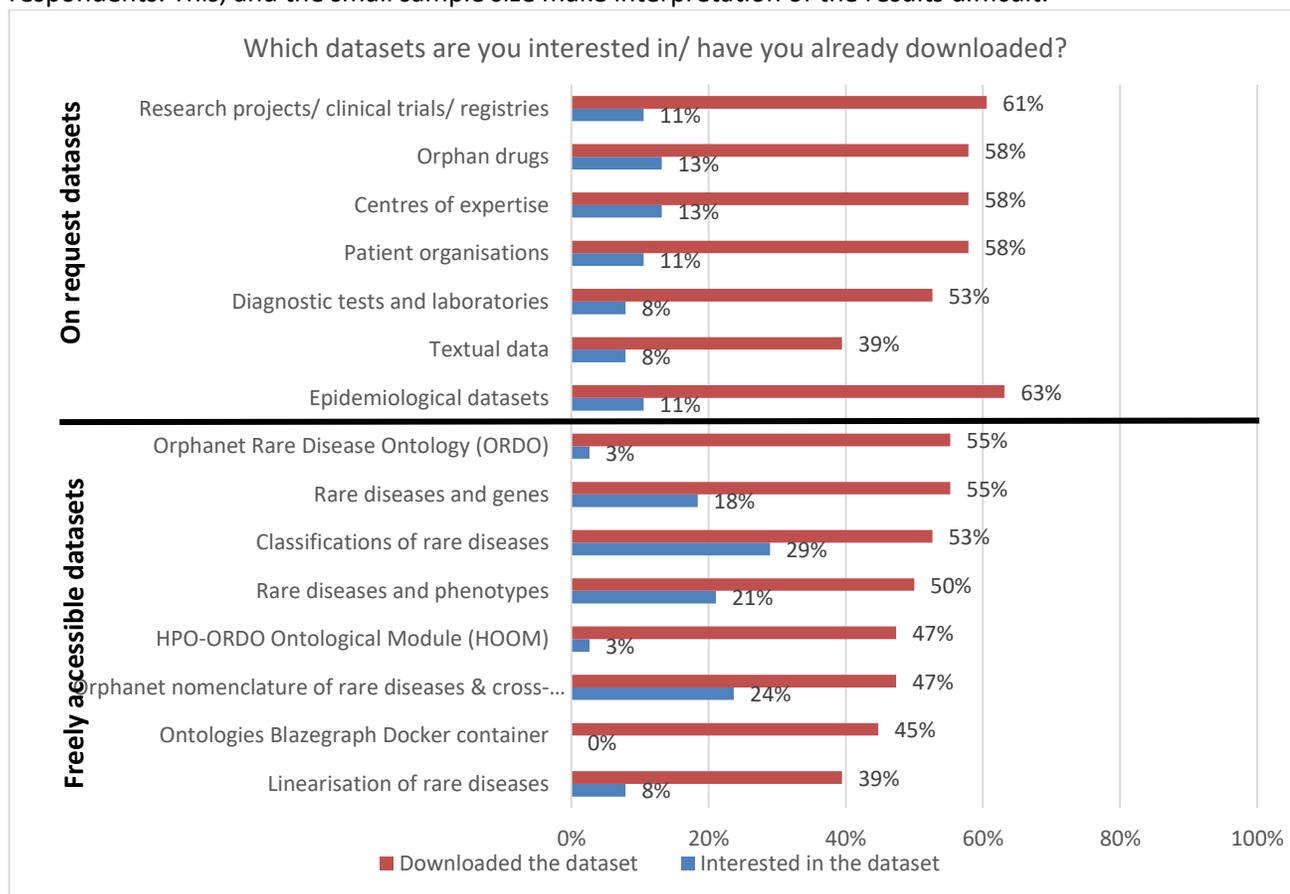

Figure 31 Interest in/ downloading of freely accessible or on request datasets (n= 38)

The results show the product most downloaded freely accessible datasets were the epidemiological dataset (63%), followed by ORDO and the rare diseases-genes datasets (55%). The classifications of rare diseases (29%) and Orphanet nomenclature of rare diseases (24%) garnered the most interest amongst the freely accessible datasets.

As regards to the on request data sets, the product most downloaded amongst the respondents was the research activities datasets (61%), followed closely by orphan drugs, centres of expertise and patient organisations (58%). Orphan drugs and centres of expertise garnered the most interest (13%).

Are you interested in/ do you use data concerning one, a few or all rare diseases? N=38

Respondents were asked to state whether they were interested principally in information concerning one specific disease, a few specific diseases, a group of diseases, or all rare diseases.

The majority of users were interested in data concerning all rare diseases (45%). We can consider that in second place came users who were interested in a selection of rare diseases, if a few or a group of rare diseases are combined (39%). Only 16% of respondents were interested in one specific disease.

What type of data formats/ accesses do you prefer? N=38

Respondents were given the possibility to indicate multiple responses to this question that aimed to find which data formats or types of access users prefer to work with.

The most preferred format was TXT (74%) followed by XML (50%) which is currently offered. The other options (SMILES/SDF, API, SPARQL, OWL, GithuB, JSON, OBO, EDF, Turtle, PDF, YAML, ODS, XLS, MDN) garnered less than 13% but showed that there was interest for SMILES/SDF and an API, not currently offered.

Do you know that we provide on request data samples and XSD/JPEG representations of the datasets? N=38

This question aimed to ascertain whether users were aware of two new features of the Orphadata website, made available in October 2018, namely the possibility to download samples of on request data, and the availability of representations of the datasets. Users were asked to indicate yes or no.

The results show that these new features were not well known, as only 32% of respondents knew of the availability of samples, and only 24% of users knew of the availability of the schematic representations of the datasets. More efforts are needed to make users aware of the availability of these recently added features.

How likely are you to recommend Orphadata to a friend or colleague? N=38

This question aimed to ascertain how likely the respondent was to recommend Orphadata to a friend or colleague by asking them to give a score of 0 to 10 (1 being not at all likely, 10 being very likely).

The average response was 8.36 out of ten and the net promoter score 44

What could Orphadata do to better serve your needs? N=38

This open question was intended to gather feedback on how the service could be improved to better serve users. Users asked for improved documentation to aid them in their use of the resources, and an easier to use site. The Orphadata website was remodelled at the end of 2018 it is hoped that this site is now clearer, but additional efforts are planned to improve the documentation available to better explain the available datasets. To note we recently added a bunch of new document in order to help users exploit our data. For instance, we added a description of our free access products, the XSD (XML schema definition) and JPEG representations of each product are also available on the site. A few users requested more frequent updates, although it was unclear whether they wished to see more frequent than monthly updates of datasets, or whether they felt that the data themselves were not up to date. It was surprising to learn that patients use this product and wish to have additional guidance on how to exploit the data.