



A BRIEF GUIDE FOR RARE DISEASE PATIENT ORGANISATIONS ON HOW TO LISTEN TO THE COMMUNITY



SPEAK UP



FOLLOW-UP



LISTEN UP



JULY 2022

Table of Contents

Acknowledgements	2
What is this Guide for?	2
STEP 1: Define Your Objectives	4
STEP 2: Assemble Your Team.	5
STEP 3: Do Your Background Research	6
STEP 4: Define Your Target Audience and Plan Your Recruitment Strategy	8
STEP 5: Select Your Method	10
STEP 6: Analyse Your Data	12
STEP 7: Disseminate Your Results	14
STEP 8: Put Your Data into Action	16
1. Co-creating rare disease Care Pathways	17
2. Developing Patient Journeys	18
3. Measuring patient experience with care	19
4. Creating a Patient-Reported Outcome Measure	19
References	20

Acknowledgements

This how-to-guide was co-developed with the rare disease patient community. EURORDIS wishes to thank the **European Patient Advocacy Group (ePAG) Steering Committee members – Angela Leburgue, Charissa Frank and Diana Marinello** for their input and dedication to the development of this guide.

What is this Guide for?

Listening to the patient community, analysing their feedback and acting on it may (i) ensure that decisions around care and design of healthcare services are needs-led [1] **(ii)** help design better services and care pathways, as there is a strong link between people having positive experiences of care and other aspects of quality, including clinical effectiveness and patient safety [2–5].

This guide aims to **support patient organisations** to understand how to best capture and act on the insights of the rare disease patient community, to ensure that the voice and lived experience of people living with a rare disease are at the heart of what patient organisations do.

You (rare disease patient organisations and patient representatives) can play a central role in this regard, i.e., by capturing the patient community needs and experience and relaying these findings to clinicians, hospital managers and decision-makers.

But first, you need to: **1)** choose the best approach and method to hear from your community, **2)** have a basic understanding on how to analyse the received feedback, and **3)** plan an effective dissemination strategy of the findings.

In addition, it will be important to estimate the costs associated with your “listening” exercise, secure funding and ensure legal and ethical compliance for all your data processing activities. While these two aspects are critical, they fall outside the scope of this guide.

**FOR A SNAPSHOT OF THIS GUIDE, AND CONCRETE TIPS AND ADVICE
YOU MAY CHECK THIS SUMMARY.**

**TO HELP YOU PLAN YOUR PROJECT AND KEEP TRACK OF YOUR DECISIONS,
HAVE A LOOK AT AND DOWNLOAD 'YOUR PROJECT PLANNER'.**

GUIDE STRUCTURE





STEP 1: Define Your Objectives

Once you have selected the topic on which you are going to focus, you need to identify the objectives of your project, i.e., **to establish the purpose and intent of your project**, what you want to achieve in broad terms.

For instance, your objective may be to *set your organisation's priorities for the upcoming months or years*, to *propose policy options to improve the diagnosis of your disease* or to *improve access to treatments for your patient group*.

Once you have identified your overall objective you may start to **reflect on what information you need to get there**, i.e., how the survey will help you achieve your objective. For instance, if your objective is to *improve access to treatments for your patient community*, you may want to:

- Have a comprehensive understanding of the types of treatments that patients are taking or have taken previously.
- Determine all treatments that have been brought to market for the specific condition(s) of the patients.
- Measure their satisfaction with current and past treatments.
- Identify their unmet needs (accessing a specific treatment, having a treatment plan...)
- Understand inequalities in access to treatment within your patient community.

Defining your overall objective and identifying what type of information you need to gather is crucial for you to: **(i) select team members (STEP 2)**; **(ii) define your target audience** (who do you want to hear from and how will you recruit them) **(STEP 4)**; **(iii) choose your data collection method** (e.g. survey, focus groups, interviews or a combination of methods) **(STEP 5)**; **(iv) set up your project's timeline**; **(v) make your data actionable (STEP 8)**, i.e. to help you plan and decide what you will do with your data and finding



STEP 2: Assemble Your Team

When assembling your team, it is good to think about how to identify and engage with all potential partners and try to involve them in the very **early stages** of the **project**. Evidence shows that involving people from the start of the project (e.g. *topic selection, definition and project design*) makes them far more likely to act on the results [6]. Gathering a **multidisciplinary team**, with varying and complementary expertise and experience, is the best way to ensure that you will have all the relevant expertise and perspectives needed to reach your objective(s).

Some of the team members you may want to integrate in your team include:

- Researchers (social scientists, statisticians)
- European Reference Network Coordinators, project managers and healthcare professionals
- Ethicists
- Patient representatives
- Stakeholders involved in or interested by the subject you want to investigate
- Research institutes or companies that may help you with parts of or all your study

Even though it is important to involve all team members as early as possible, you should allow for some **flexibility**, as sometimes it will not be possible to recruit and integrate all partners at the same point in time.



STEP 3: Do Your Background Research

Before starting to develop the approach to capture the voice of your community, it is essential that you conduct some **background research**. Putting some effort into performing background research will allow you to:

- **Identify best practices**, avoid “reinventing the wheel” and identify whether the data that you are trying to collect already exists.
- **Gather relevant information to better frame your listening exercise** and guide your data analysis, for instance by comparing your findings with existing results. Ensure that your project builds on and goes beyond what is already known!
- **Help identify experts** whom you can contact to partner or seek advice (e.g. authors of scientific papers).
- **Get inspired by similar initiatives** in other disease areas or communities and identify approaches, methodologies, or ideas that you can adapt to your own project.

Where can you look for reliable information? These are all good places to start:

- **Scientific Literature:** you may search in medical literature databases, such as [PubMed](#) or [Google Scholar](#).
- **Grey literature:** this refers to non-medical or scientific literature, e.g. guides published by patient organisations, medical societies or reliable national or international organisations (governments, OECD, WHO, etc.).
- **Existing databases:** [Proqolid](#) (clinical outcomes assessment measures)¹; [EORTC quality of life library](#) (quality of life measures); [PROMIS database NIH](#) (patient reported outcome measures repository); [UK Service for economic, social and population data](#); [National Center for Health Statistics](#), USA (includes questionnaires, data sets and other documentation on various health topics); [Qualitative data repository](#), USA (stores qualitative and multi-method research in the social sciences and related disciplines). You can find a comprehensive list of data portals focusing on different health-related areas and hosting datasets, as well as other documents from around the world [here](#).
- **Previous, current or upcoming [Rare Barometer surveys](#):**
 - **Global results** are available online, on the Rare Barometer website.
 - **Specific results:** patient organisations who are members of EURORDIS can ask for results of studies involving their own disease area, if there are enough respondents (per disease, disease group, country, geographical area, etc.). Disseminating Rare Barometer surveys will increase your chances of having enough respondents and access to your results!
 - **Using questions from past surveys:** you can also ask for the questions of previous surveys and use those questions in your own questionnaire. You can find the questions in the [Rare Barometer question repository](#), or send an email to rare.barometer@eurordis.org

¹ Rare disease patient organisations can obtain full access to PROQOLID until 2024 (i.e. for the duration of the ERICA project). To obtain full access, contact Céline Desvignes at: Celine.Desvignes-Gleizes@mapi-trust.org

- **Experts in the topic that you are planning to work on can also be a highly useful source of information.** Reach out to and brainstorm with clinicians, other patient representatives and patient organisations, medical and scientific societies, universities and research institutes/groups.

ADDITIONAL RESOURCES

- The [Rare Barometer question repository](#) has questions in 23 languages that you may directly use or adapt to your context and needs. The repository includes a list of questions, items and instructions, organised by variables such as sociodemographic information, among others.
- A helpful resource is this short guide by NHS England that provides an overview of the different types of information already available and brief explanations of how they can be used: [Insight – What is already available?](#)



STEP 4: Define Your Target Audience and Plan Your Recruitment Strategy

Defining your target audience should be straightforward, but it is important to think carefully about who you need to hear from. To do this you should be clear about your **objective and what type of information you need to gather to achieve it (STEP 1)**.

- If you want to find about the healthcare preferences of people living with Duchenne Muscular Dystrophy in Europe, then your sample needs to reflect this (i.e., the participants you will be recruiting).
- If you are interested in learning about care pathways in rare epilepsies in Spain, then focus on the groups experiencing them.
- In some cases, it may not be adequate or possible for patients to participate or answer for themselves (e.g. when most patients are children, or you want to explore the burden of caregivers). In these situations, family caregivers or family members would be your target audience.

After clearly determining who your target audience is, you can plan your **recruitment strategy** and choose the **dissemination channels** you are going to use to recruit your potential respondents. It is also important to identify the partners who can help you maximise the effectiveness of your recruitment plan.

Some examples of possible dissemination channels and potential partners include:

DISSEMINATION CHANNELS AND POTENTIAL PARTNERS FOR RECRUITMENT

DISSEMINATION CHANNELS	POTENTIAL PARTNERS
Social media, newsletters, websites, online patient communities (e.g. RareConnect) Conferences, webinars, workshops Emails, letters	Patient organisations, patient representatives, clinical leads, medical societies, clinicians, health authorities, European Reference Networks, European Patient Advocacy Groups
Direct recruitment at hospitals via flyers and posters	Clinicians and hospital managers

TIPS

- **Know where to find your target audience.** You are often the best one to know the most popular social media channels in your community. This allows you to tailor your recruitment strategies to your target audience preferences to maximise your reach and, hopefully, optimise your time and resources. Keep in mind that some of your target audience may have characteristics (e.g. age, visual impairment, etc.) that greatly impact their communication and social media outlet habits.
- **Challenge the “hard-to-reach” groups concept.** Difficulties in recruiting under-represented population groups may stem from not using the right channels and methodologies to reach out to them, e.g. for elder patients consider asking clinicians to print a survey and distribute it to patients during their appointments.
- **Be aware that your data collection approach may influence the choice of your recruitment strategy.** If you are using an electronic or online survey, then using social media and/or emailing are probably the most suitable options. On the other hand, if you are planning to conduct one-on-one interviews or focus groups with a specific group of people being seen at a given clinic, then conducting direct recruitment at the clinic (maybe in combination with invitation letters or emails) is probably your best option.
- **Diversify your recruitment channels and partners to increase the number of respondents and avoid bias.** Often a combination of multiple channels and involvement of different partners is the best approach (e.g. recruiting only patients who are in contact with patient organisations).
- **Adapt the number of respondents to your population and to your needs** (and adapt your analysis to the number of respondents – (see [STEP 6](#)):
 - For **qualitative research**, people are usually interviewed “until saturation” is reached, i.e., until you stop learning anything new from the interviewees regarding your topic. Depending on your research questions, on your level of expertise on the topic and on the size of your population, this number can be very different (from around 10 to 70 people or more).
 - For **questionnaires**, the sample size depends on your population size, and on the degree of precision you wish to have in your findings. Several tools exist on the internet, which allow you to have an idea of the ideal sample size for your project (you can find one [here](#)). Please note that these tools are not always adapted to small samples. In general, it is usually good to have at least 30 respondents, but you will have to be mindful of the margin of error² of your results, which you can calculate [here](#), for instance. Rule of thumb: the larger your sample, the more accurate your results will be. If you do have a large population, however, your results will no longer change after reaching 1000 respondents – additional answers will only allow you to dig more into sub-populations (for instance: results for carers and for patients; for men and for women; for different subtypes of one disease...).
- **Be strategic and seize the most adequate time window to launch your survey and keep it open for participation.** In general, certain periods during the year, such as Christmas or the summer months (especially July and August), should be avoided, as participation tends to be lower. Regarding the question of ‘**How long should I keep my project open for participation?**’ published studies of online questionnaires in rare diseases reported that most projects had an active recruitment/participation period lasting 1 to 6 months with only two out of 44 cases—one of them a pilot study—recruiting for under a month [7]. However, the length of this period can largely vary, depending on various factors, including the characteristics of your survey (e.g. number of questions, type of data collection method), your objectives, and your target population.

² Margin of error is a range of values that differ from the actual survey results.



STEP 5: Select Your Method

Which approaches are the most appropriate to capture the voice of your community? The most honest answer to this question is: **it really depends!** People's insight and feedback about their experiences, needs and priorities related to health systems and healthcare can be gathered in a wide range of ways, and there is **no one-size-fits-all approach**.

The **key questions** to answer to determine the selection of your methodology are:

- What are your specific objectives? Why are you collecting your community views on a given topic? Try to be specific on the purpose ([STEP 1](#)).
- Would your target population feel more comfortable and/or be more likely to participate in an online survey, an interview, an in-person focus group? Are there any barriers linked to language, sensory loss, geography, or other issues that you should consider? If yes, how can you remove these barriers? ([STEP 4](#))
- How will the information you gather be used; how will you action your data ([STEP 8](#))?

There are different approaches and methods that you can use, both alone or in combination, to listen to and learn from your community. Broadly, we can divide these methods into **3 main categories**:

- 1. Quantitative methods:** Quantitative methods' results are expressed in "numbers" and data are commonly collected through questionnaires. You can find some inspiration for your **questionnaire**, including questions you can adopt and adapt to your project, in the [Rare Barometer question repository](#).
- 2. Qualitative methods:** The main goal of qualitative methods is to understand processes, actions or opinions in ways that are more interactive and attentive to individual behaviours. Through qualitative approaches you may collect "stories" through **interviews** or **group discussions**; but also "facts" through **direct observation**, for instance. Stories collected through qualitative methods can humanise your findings, illustrating them in a more personal way. They can also provide more detailed insights into people's perspectives on what is being surveyed; what works well and what could be improved. If your main goal is to collect figures, but you still want to include some quotes from respondents, you can also include **open-ended questions** in a questionnaire.
- 3. Mixed methods:** This approach combines several types of methods, quantitative or qualitative. Mixed methods can be a powerful tool to maximise and guide data collection and analysis. For instance, when designing a questionnaire, conducting first interviews or focus groups can help identify what matters most to patients or carers, so that you do not include only your own conception of what is important. Rare Barometer adopted this approach for its survey on the [Journey to Diagnosis for People Living with Rare Diseases](#). Prior to the launch of the survey, patient representatives participated in an online panel to exchange views and share experiences on obtaining a diagnosis. This was followed by eight individual interviews. The findings were used to guide the development of the questionnaire, with the support of a committee composed of experts in the field of diagnosis, including policy experts, patient advocacy organisations, sociologists, and corporate partners. Conducting interviews or focus groups can also be useful to better understand and interpret the results of a questionnaire.

TIPS

- **Download [your project planner](#) and this [summary table](#)** to help you choose your data collection method and tools, and to get concrete tips and advice.
- **Test the tools** (e.g. questionnaire, interview guide) by **conducting a small pilot** to ensure usability, understandability and content appropriateness (e.g. interview rehearsal before sharing with participants). By doing so you can solve any technical or methodological limitations and bugs, ensure the tool is tailored to your target population, and make sure the language you are using is understandable.
- **Formulate good questions.** View some good tips [here!](#)
- **Translate as much as possible.** Translating your survey and associated materials such as communication material, facilitates participant engagement. In turn, the number of participants impacts the robustness of your results. If you have an international team, it can be interesting to translate the questionnaire in one or two languages before finalising it to improve the clarity and precision of your questions. Inaccurate translations will have a detrimental impact on the quality of the results, don't risk it! To avoid this, make sure you check and validate the translations:
 - When the topic you are exploring is medical or has medical jargon, have a (bio)medical translator validating the translations to make sure questions, scales and results are comparable and reliable.
 - When translating, be aware of "false friends". Using synonyms and translating them back helps to identify the right word.
- **Carefully organise and manage your data collection:** save a copy of your questionnaire, or of the different versions of your guides for qualitative methods (interviews, focus groups, observation...); monitor responses regularly (number of respondents, response rate, completion rate) to adapt your dissemination strategy accordingly; gather all your data (e.g. filled questionnaires, interview guides, interview recordings, transcripts, notes) and save it in a secure space accessible only to the research team.

ADDITIONAL RESOURCES

- [Video tutorial](#) offering tips on how to choose your data collection method.
- [Summary](#) providing more information, practical guidance and advice on the data collection methods and their associated tools.
- Helpful, bite-size guides by NHS England on: 1) [Writing an effective Questionnaire](#) and 2) [Building greater insight through Qualitative Research](#).



STEP 6: Analyse Your Data

How do you analyse and interpret the information you gather? You should plan early on **how, when and by who** your survey results will be analysed and interpreted. Reflecting on your data analysis in advance will allow to:

- **Secure external expertise if needed** in case your organisation lacks the necessary skills in-house.
- **Assist in developing your data collection method (STEP 5)**. Thinking about how you are going to analyse and interpret the information you aim to gather will help you to: **1)** reflect on what data is meaningful and **2)** decide how you will structure your data collection, the format and order of your questions. In the end, you will create a more concise and informative data collection instrument.
- **Organise your budget and resources**. Results analysis may not only be time-consuming but can also be expensive. Estimating these costs ahead of time, will allow you to devise alternative strategies to avoid a scenario where you have gathered the data but have no one or no resources to properly analyse them.

Whether you are going to work with numbers (quantitative data) or stories (qualitative data) will determine your data analysis method.

Quantitative data analysis. There are several steps that you must follow when performing this type of analysis:

- 1. Check for data completeness.** Verify that all the surveys you received are complete, i.e. that all questions (or all mandatory/ essential questions) are answered.
- 2. Check for duplicated answers.** Sometimes the same person may complete the survey more than once.
- 3. Establish inclusion and exclusion criteria.** These criteria are fundamental and refer to the characteristics that participants must have to be included (inclusion criteria) or excluded (exclusion criteria) from a study. For example, you may decide to exclude incomplete surveys, respondents with specific diseases, or who completed the survey too fast. Establishing inclusion and exclusion criteria ensures that the data you analyse are high-quality and relevant to your project aims.
- 4. Perform statistical analyses.** Analysing quantitative data requires running statistical analyses, there is no getting around it. Choosing the correct statistical test(s) to apply to your data should be done with the support of an expert. There are some basic statistical test(s) that you can run to get an initial broad understanding of your results, such as calculating percentages (associated with the frequency that something is present in our data, for example, the percentage of family carers who replied to the survey in relation to the total number of respondents), and averages or medians (e.g. the median age of the respondents). When analysing your data, you should always take your sample size into account (see [STEP 4](#)).

Qualitative data analysis. Thematic analysis is possibly the most popular type of qualitative data analysis. [This paper provides](#) a good example of the full process of conducting a rigorous thematic analysis [8]. There are broadly 3 different stages for conducting a thematic qualitative analysis:

- 1. Organise the data.** Convert the data into a usable format and structure, such as transcribing interviews/focus groups audio recordings into written text.

2. **Explore and familiarise yourself with the data.** Read through the data and begin to identify potential themes, i.e. those features in the participants' accounts that characterise specific perceptions or experiences relevant to the research question.
3. **Code and classify.** Themes should then be organised by developing a coding system. See an illustration of thematic coding in the example below:

CODING EXAMPLE

Interview extract	Codes
<p>Personally, I'm not sure. I think the climate is changing, sure, but I don't know why or how. People say you should trust the experts, but who's to say they don't have their own reasons for pushing this narrative? I'm not saying they're wrong, I'm just saying there's reasons not to 100% trust them. The facts keep changing – it used to be called global warming.</p>	<ul style="list-style-type: none"> • Uncertainty • Acknowledgement of climate change • Distrust of experts • Changing terminology

Source: <https://www.scribbr.com/methodology/thematic-analysis/>

It is possible to conduct this kind of qualitative data analysis manually, using a Word document or an Excel spreadsheet. However, there are a number of helpful software packages available to analyse qualitative data (see the [additional resources](#) below).

Once you have analysed your data, it is time to interpret and develop possible explanations for your findings. Ask yourself **“What can these results mean?”**. Results interpretation may not be a straightforward exercise and you should be very careful to avoid over-interpretation. It is better to state that you cannot yet explain the full meaning of the results, propose hypothetical explanations, or state that further research is needed, rather than over-interpret results.

TIPS

When difficulties or doubts arise:

- **Put your results into context**, framing the results within the context of your research aim or target audience. In certain cases, if possible, reaching back to the participant(s) can be helpful.
- **Discuss the results with your team.**
- **Check if published information exists** that can help you better understand and frame your results.

ADDITIONAL RESOURCES

- A selection of [data collection and analysis tools](#) for your project.
- [Video tutorial](#) on quantitative data analysis.
- [Video tutorial](#) on simple qualitative data analysis.



STEP 7: Disseminate Your Results

Remember that listening to your community is just half of the process. The feedback you have gathered and processed needs to reach the people who will use the information to take decisions either at personal, healthcare provider or policy level.

First, share the results with your community! You will build trust as you give back and keep your community engaged and informed. Next, share your results with decision-makers and other stakeholders.

Sharing the methods, limitations and findings of your research is absolutely essential and will allow others to use and build on what you have developed. Always remember to **be clear about the methodological limitations** of your research and **highlight any biases** that your data might have. Discuss all this carefully with the person in charge of the data analysis.

Devising clear, preferably multi-channel, multi-format (e.g. video, podcasts, presentations) and multi-language results dissemination strategies should be a top priority from the outset. The table below provides some suggestions on communication channels and formats.

COMMUNICATION CHANNELS AND FORMATS

COMMUNICATION CHANNELS	COMMUNICATION FORMATS
Conferences and other gatherings	Project Summary Reports
Social media and websites	Video
Scientific journals	Audio (e.g. podcast)
Media (e.g. magazines, TV)	Infographics/posters
Email campaigns	Presentations
Newsletters	Articles, posts
	Scientific papers
	Factsheets

You can combine different communication channels and pair them with an array of formats. Your motto should be **'communicate the right message, in the right format, through the right channels to the right people at the right time'**.

TIPS

- **Plan and devise your result dissemination strategies in advance.** You should begin to define your dissemination strategies since the beginning of the project, including anticipated target audiences, materials you will need to create (e.g. social media posts, posters, etc) and a potential timeline for all the result dissemination activities you aim to do.
- **Periodically communicate your findings to your community.** Sharing intermediate results and giving periodic updates builds greater trust, promotes transparency and engagement, maximising the community endorsement of your findings. It is also an opportunity to obtain additional feedback and guidance from your community if you wish to do so. However, sharing intermediate results can be tricky. You may fear creating too many expectations or giving too much away too soon – but finding a good balance is possible!
- **Determine how you would like to present your results.** You may choose several formats to present your results: graphs, tables, schemes and other figures, such as word clouds. This can be decided based upon: **1) the type of data you have** (quantitative or qualitative); **2) how and to whom you are going to present your data** (e.g. publish in a scientific paper; present your results at a conference; share them in a social media post).
- **Make information human.** Storytelling using slides or videos (including quotes, photos, etc.) will generate greater empathy, connectivity and engagement.
- If you decide to publish your results in a scientific journal, **make sure the journal is [open access](#)**. Likewise, create all your materials as much as possible under the [Creative Commons Licence](#) (CC), so that everyone can reuse them.



STEP 8: Put Your Data into Action

Healthcare is to a great extent a relational experience, and as such, **connecting and listening to patients' needs** is important to design responsive healthcare services.



THE INSIGHTS FROM PERSONS LIVING WITH A RARE DISEASE SHOULD BE CENTRAL TO THE PLANNING, ORGANISATION AND EVALUATION OF HEALTHCARE SERVICES.

The 4 projects described in this section illustrate how you can turn your data into action. Each project has used a different approach to capture the voices of the rare disease patient community and the results have been used in different ways.

1. CO-CREATING RARE DISEASE CARE PATHWAYS

Care pathways are comprehensive and complex plans that guide and support the organisation of care for a well-defined group of patients during a well-defined period [9]. They can have different degrees of granularity and can be multi-professional, typically incorporating multiple guidelines [10]. They provide a compass for all the persons involved in the organisation and delivery of care, including patients and carers. This enables coordination between different levels of care - hospital, community, social and primary care- and supports the delivery of integrated care.

Care pathways for coordinated care in rare diseases are largely missing [11]. Recently two different initiatives by [Irish National Rare Diseases Office](#) (NRDO) and ERN [ReCONNET](#) have proposed methodological approaches to develop rare disease care pathways [12,13]. Both methodological approaches included the participation of patients as co-designers of the pathways.

In the first case, NRDO shared the relevant care pathway document with a patient representative who was asked to invite other patient advocates to attend an online workshop jointly led by [Rare Diseases Ireland](#) and NRDO. The main objective of this workshop was to listen to the rare disease patient community, allowing NRDO to:

- capture the patients' experiential knowledge;
- better understand the common needs and interventions which patients with rare diseases prioritise;
- enhance the relevance and utility of the care pathways.

Due to the diverse operational structures of the patient organisations involved, a range of methods were used to listen to the patient community: patient representative co-ordinators out-reach within their networks; establishment of short-term working groups; review by patient organisation boards; and direct liaison of patient representatives with clinical leads [12].

In the second case, ERN ReCONNET has defined a methodology to develop “**reference model**” pathways for rare and complex conditions [13]. Specifically, the first phase of the proposed methodology includes the collection of patient views and perspectives through a survey based on the principles of narrative medicine [14] that should be co-designed in English by patients affected by the relevant disease. Once the survey is finalised, it should then be translated into different EU languages, if possible, involving patients and patients' representatives from the different countries in the validation process.

The survey to capture patients' perspectives through this methodology would consist of the following sections:

1. an introduction to describe the scope of the survey and explain the eventual further use of the stories,
2. demographic questions to capture a profile of the respondents,
3. a free-text space dedicated to writing the stories (qualitative data, 3600–5000 characters) with a set of questions to support and inspire the patient telling the story.

2. DEVELOPING PATIENT JOURNEYS

Patient journeys are personal testimonies from people with first-hand experience of living with a rare disease. They map the needs of patients along the different stages of their journey, from first symptoms, diagnosis, to treatment and follow-up, through the eyes of the patients or carers [15].

EURORDIS provided an [open-ended questionnaire](#) to collect the information for the patient journeys and [legal guidance to collect the data](#) in Belgium, The Netherlands, Spain, Portugal Germany and France. The questionnaire was organised into **3 levels: 1) clinical presentation, 2) challenges and needs identified by patients, and 3) their expectations on how care services should improve**. Patient representatives reached out to their networks to fill out the questionnaire based upon their experience, either using an online survey platform or via interviews. The information collected for the patient journeys was usually reviewed by clinicians and was summarised in a graphic to support user-friendly communication (you may access the visual template [here](#)).

Several ERNs have used this tool to capture the needs of the rare disease patient community under their scope.

EUROPEAN REFERENCE NETWORK	RARE DISEASE/COMPLEX CONDITION
ERN-RND	<ul style="list-style-type: none"> • Huntington's disease • Friedreich's Ataxia
ERN SKIN	Patient journeys for various rare skin diseases are available here .
ERN GENTURIS	Patient journeys for various genetic tumour risk syndromes are available here .
ERN CRANIO	Patient journeys for various rare and/or complex craniofacial anomalies and ear, nose and throat (ENT) disorders are available here .
ERN ITHACA	<ul style="list-style-type: none"> • Rett Syndrome • Williams Syndrome • Prader Willi Syndrome • Spina Bifida • Pitt-Hopkins Syndrome • Patient Journey Common Needs
ERN LUNG	Patient journeys for various rare lung diseases are available here .
ERN ERKNet	Patient journeys for various rare kidney diseases are available here .
ERN euROGEN	<ul style="list-style-type: none"> • Interstitial cystitis • Anorectal malformation • Bladder Exstrophy patient journey
EpiCARE	<ul style="list-style-type: none"> • Dravet syndrome • Hypothalamic Hamartoma • Alternating Hemiplegia of Childhood patient journey

In some ERNs, patient journeys have allowed patient representatives and clinicians to engage in productive discussions around patients' perspective on gaps in care pathways, common unmet needs, standards of care, etc. As such, patient journeys can be an effective and simple tool for patient representatives to capture and convey the needs of the rare disease patient community specifically around healthcare services, allied therapies, and psychological support.

3. MEASURING PATIENT EXPERIENCE WITH CARE

The [H-CARE project](#) was initiated by four ERNs ([GENTURIS](#), [eUROGEN](#), [ERN-LUNG](#) and [ERKNet](#)) and supported by EURORDIS. It aims to develop a common feedback mechanism that could be used by healthcare providers across different EU countries to systematically collect data on patient experience with care.

By measuring patients' and carers' experience of health care, the H-CARE project seeks to understand **1)** how to improve medical care to address the needs of people living with a rare disease and **2)** how multidisciplinary care teams specialised in the treatment of rare diseases could help to reach this goal, ultimately improving health outcomes [16].

The project started with a pilot survey (2019-2020) that tested the feasibility of a feedback mechanism. Since there is no validated questionnaire to measure experience with care for rare and complex diseases, the project partners decided to use the PACIC-S, a scale validated for common chronic diseases [17]. The 11 items of the PACIC-S were adapted for patients and carers living with rare or complex diseases, and for specialised care. These adapted versions can be found [here](#) and [here](#). You can also access the [key findings](#) and the [detailed results](#).

The pilot survey revealed certain limitations with the scales used and a need to develop specific instruments to measure the care experience of rare disease patients and carers. Going forward, the project now aims to validate two **Patient Reported Experience Measures (PREMs)** in several languages, and to use these questionnaires as a monitoring indicator for the patient-centeredness of the ERNs and their members.

4. CREATING A PATIENT-REPORTED OUTCOME MEASURE

Patient-Reported Outcome Measures (PROMs) are questionnaires that capture a person's perception of their own health. PROMs can measure quality of life, daily functioning, symptoms, and/or other aspects of their health and well-being [18].

The Duchenne muscular dystrophy patient community was heavily involved in the development of a PROM to better understand patient and caregiver perception of abilities related to upper limb function in daily life, such as feeding, washing, and leisure activities [19].

The opinions of patients and their families were heard throughout the different stages of the questionnaire development by soliciting their views on task difficulty and relevance. Different methods were used to capture their actionable input, ranging from **focus groups** to **pilot testing of the questionnaire** to assess its adequacy and suitability. Their involvement ensured that the PROM is not only disease- and population-specific but also assesses clinically meaningful upper limb function in this population.

References

1. Bolz-Johnson M, Kenny T, Le Cam Y, Hernando I. Our greatest untapped resource: our patients. *J Community Genet.* 2021;12: 241–246. doi:10.1007/s12687-021-00524-5
2. Doyle C, Lennox L, Bell D. A systematic review of evidence on the links between patient experience and clinical safety and effectiveness. *BMJ Open.* 2013;3: 1570. doi:10.1136/bmjopen-2012
3. Cornman DH, White CM. Discerning the Perception and Impact of Patients Involved in Evidence-based Practice Center Key Informant Interviews. *AHRQ Methods for Effective Health Care.* 2017 Sep. doi:10.23970/AHQEPCWHITEPAPER1
4. Fønhus MS, Dalsbø TK, Johansen M, Fretheim A, Skirbekk H, Flottorp SA. Patient-mediated interventions to improve professional practice. *Cochrane Database of Systematic Reviews.* John Wiley and Sons Ltd; 2018. doi:10.1002/14651858.CD012472.pub2
5. Sharma AE, Knox M, Mleczko VL, Olayiwola JN. The impact of patient advisors on healthcare outcomes: A systematic review. *BMC Health Serv Res.* 2017;17. doi:10.1186/s12913-017-2630-4
6. de Silva D. *Measuring patient experience.* London; 2013.
7. Francisco R, Brasil S, Pascoal C, Jaeken J, Liddle M, Videira PA, et al. The road to successful people-centric research in rare diseases: the web-based case study of the Immunology and Congenital Disorders of Glycosylation questionnaire (ImmunoCDGO). *Orphanet J Rare Dis.* 2022;17. doi:10.1186/s13023-022-02286-w
8. Nowell LS, Norris JM, White DE, Moules NJ. Thematic Analysis: Striving to Meet the Trustworthiness Criteria. *Int J Qual Methods.* 2017;16. doi:10.1177/1609406917733847
9. Vanhaecht K, Bollmann M, Bower' K, Gallagher C, Gardini A, Jen Guezo ' L, et al. Prevalence and use of clinical pathways in 23 countries-an international survey by the European Pathway Association. *Int J Integr Care.* 2006. Available: www.E-P-A.org
10. Lawal AK, Rotter T, Kinsman L, Machotta A, Ronellenfitsch U, Scott SD, et al. What is a clinical pathway? Refinement of an operational definition to identify clinical pathway studies for a Cochrane systematic review. *BMC Medicine.* 2016;14. doi:10.1186/s12916-016-0580-z
11. Walton H, Hudson E, Simpson A, Ramsay AIG, Kai J, Morris S, et al. Defining coordinated care for people with rare conditions: A scoping review. *Int J Integr Care.* 2020;20: 1–16. doi:10.5334/IJIC.5464
12. Ward AJ, Murphy D, Marron R, McGrath V, Bolz-Johnson M, Cullen W, et al. Designing rare disease care pathways in the Republic of Ireland: a co-operative model. *Orphanet J Rare Dis.* 2022;17: 162. doi:10.1186/s13023-022-02309-6
13. Talarico R, Cannizzo S, Lorenzoni V, Marinello D, Palla I, Pirri S, et al. RarERN Path: a methodology towards the optimisation of patients' care pathways in rare and complex diseases developed within the European Reference Networks. *Orphanet J Rare Dis.* 2020;15. doi:10.1186/s13023-020-01631-1
14. Fioretti C, Mazzocco K, Riva S, Oliveri S, Masiero M, Pravettoni G. Research studies on patients' illness experience using the Narrative Medicine approach: a systematic review. *BMJ Open.* 2016;6. doi:10.1136/bmjopen-2016-011220
15. Bolz-Johnson M, Meek J, Hoogerbrugge N. "Patient Journeys": improving care by patient involvement. *Eur J Human Genet.* 2020;28: 141–143. doi:10.1038/s41431-019-0555-6
16. Rickert J. Patient-Centered Care: What It Means and How to Get There. *Health Affairs Blog.* 2012. Available: <https://www.healthaffairs.org/do/10.1377/forefront.20120124.016506>. Accessed 20 Jun 2022.
17. Gugiu PC, Coryn C, Clark R, Kuehn A. Development and evaluation of the short version of the Patient Assessment of Chronic Illness Care instrument. *Chronic Illness.* 2009;5: 268–276. doi:10.1177/1742395309348072
18. Churrua K, Pomare C, Ellis LA, Long JC, Henderson SB, Murphy LED, et al. Patient-reported outcome measures (PROMs): A review of generic and condition-specific measures and a discussion of trends and issues. *Health Expectations.* John Wiley and Sons Inc; 2021. pp. 1015–1024. doi:10.1111/hex.13254
19. Klingels K, Mayhew AG, Mazzone ES, Duong T, Decostre V, Werlauff U, et al. Development of a patient-reported outcome measure for upper limb function in Duchenne muscular dystrophy: DMD Upper Limb PROM. *Dev Med Child Neurol.* 2017;59: 224–231. doi:10.1111/dmcn.13277



EURORDIS-RARE DISEASES EUROPE

Plateforme Maladies Rares ♦ 96 rue Didot
75014 Paris ♦ France

EURORDIS BRUSSELS OFFICE

Fondation Universitaire ♦ Rue d'Egmont 11
1000 Brussels ♦ Belgium

EURORDIS BARCELONA OFFICE

Recinte Modernista Sant Pau ♦ Pabellón de Santa Apolonia
Calle Sant Antoni M^a Claret 167 ♦ 08025 Barcelona ♦ Spain

EURORDIS.ORG

This paper was produced thanks to funding received under an operating grant from the European Union's Health Programme (2014-2020).



Co-funded by
the Health Programme
of the European Union